

The GenomEthics Study

End of Project Report and Evidence of Impact and Reach, 2010-2016



Dr Anna Middleton
Principal Staff Scientist, Ethics Researcher for the DDD Study

Click Here

Ethics and Genomics Survey



- Sharing of Pertinent Findings
- Sharing of Incidental Findings
- Categorizing Incidental Findings
- Relations with Risk
- Raw data
- Duty of Genomic Researchers
- Filter of Genomic Information
- Consent for genomic research
- Last few questions about you

Sharing of Pertinent Findings

- Should Pertinent Findings from genome studies be made available to research participants?
 - Research participants should be able to receive pertinent findings if they want them
 - I don't think pertinent findings from research projects should be available
 - I don't know

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Next »

www.GenomEthics.org

Genomics.org

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Ethics and Genomics Survey

Questions about you

- Sharing of Pertinent Findings
- Sharing of Incidental Findings
- Categorizing Incidental Findings
- Relations with Risk
- Raw data
- Duty of Genomic Researchers
- Filter of Genomic Information
- Consent for genomic research
- Last few questions about you

Do you work within the healthcare profession? (including those involved directly with patients in a hospital/clinical setting)

Yes No

« Previous Next »

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Life from a Wellcome Trust perspective

What are your views on what happens to your genomic information?

FEB, 2012

by Wellcome Trust

Wellcome Trust

Tags: Bioethics, Dr Anna Middleton, Ethics, Genomics, Personal genomics, Wellcome Trust Sanger Institute



A screenshot from one of the videos

We're closer than ever to harnessing whole genome sequencing for everyday medical use. But what would you want to know – and what would you not? Anna Middleton invites you to take part in a new study looking to understand the ethical implications.

Public engagement about genomics is demanding at the moment. A whole genome test on a single saliva sample can tell a person about their genetic risks for hundreds of conditions all in one go, ranging from whether they are predisposed to developing Alzheimer's disease or diabetes through to whether they are sensitive to certain antibiotics or whether their children will have inheriting something specific.

We are a global charitable foundation dedicated to achieving extraordinary improvements in health by supporting the brightest minds. [Read more](#)

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The GenomEthics Study

Dr Anna Middleton

Principal Staff Scientist, Ethics Researcher for the DDD Study

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DDD Ethics Project

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GenomEthics Project Background

Start date: 8th November 2010

Maternity leave: Feb 2012-Jan 2013

End date: 30th September 2016

Full time work on project 08/11/10 - 01/05/15

50% work on the project 01/05/15 - 30/09/16

Total time worked on project, Dr Anna Middleton: 4 years 3 months fte

Research Brief:

Design and deliver a social sciences research study to understand what the public, research participants, patients, health professionals and genomic researchers think should happen with respect to the sharing of incidental findings from genome sequencing research studies.

The principal research question/objective

This is an exploratory study; the objectives are to investigate the following:

- Attitudes towards sharing of 'pertinent findings' from whole genome studies
- Attitudes towards sharing of 'incidental findings' from whole genome studies
- Attitudes towards receiving information relating to different categories of genomic information
- Attitudes towards the sharing of raw genomic data
- Attitudes towards genomic researchers sharing incidental findings even if this means compromising their ability to answer their research question

1. Design of the GenomeEthics study

Creation of the study

The way the ethics study was structured and designed was open. As delivered in my job interview, I suggested a mixed-methods project that would use an online survey plus series of qualitative interviews to gather data. I titled this the 'GenomEthics' study. In order to ensure that the views of the management committee and various stakeholders were represented in the research and more importantly, that the right questions would be asked, I started the research process with an extensive literature review, and used the themes from relevant literatures as a basis for exploration with all stakeholders. This culminated with the production of the Ethics and Genomics study protocol (Appendix A).

Compliance with Data Protection Legislation

As part of compliance with internal Sanger Institute policy I completed the Human Materials and Data Management Committee (HMDMC) paperwork addressing how the research data would be stored, encrypted and protected as per the Data Protection Act. This is particularly pertinent as some identifiable data (Level 4) would be collected from research participants and since this level of data is not routinely held on campus, new pathways needed to be developed, this involved discussion and agreement from David Davison (then COO), Tim Hubbard and Paul Bevan. The HMDMC paperwork has been adjusted several times in line with an evolution of thinking on campus and the latest version of this can be viewed in Appendix B.

Creation of a Lone Worker Policy

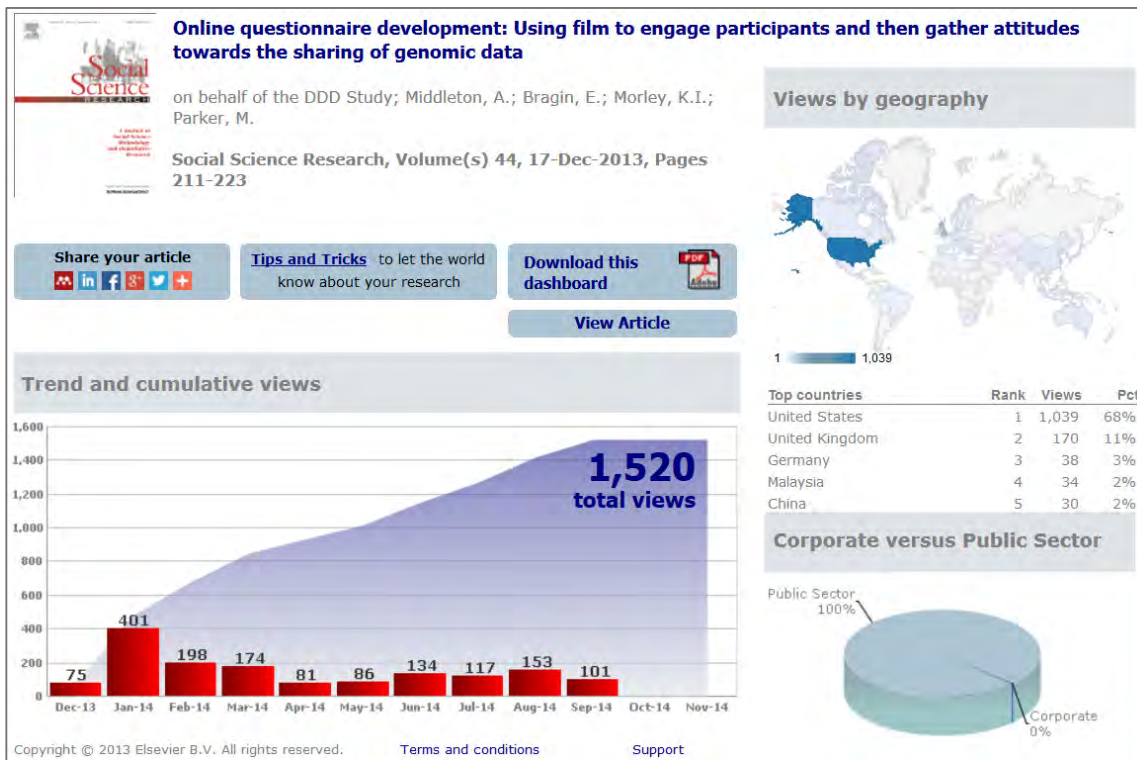
In the early stages of the project the intention was to interview research participants off campus. In order to comply with Sanger Institute Health and Safety requirements I created a Lone Worker Policy, which has been risk assessed and endorsed by the Health and Safety team and now adopted as Sanger policy. The aim of this piece of work was to ensure sensible steps were taken to protect myself or any interviewer working in a research setting with members of the public in their own homes or off campus (see Appendix C).

REC approval

After the creation of the study protocol, interview schedule, consent form, participant information sheet and draft survey, in July 2011 I submitted the REC paperwork and received favourable REC approval in August 2011 (Appendix D).

Design of the Online Survey

The creation of the online survey took 9 months and 19 iterations. This involved discussion of content with all stakeholder groups (lay members of the public, parents of children involved in the DDD project, genetic health professionals, clinical scientists, genomic researchers, DDD management committee and Sanger ethics and policy staff). The discussion took a structured format and consisted of a focus group, 4 face validity tests and 5 pilot studies. I also conducted a readability test and reliability testing, to ensure that the bespoke survey was as robustly designed as possible. Appendix E shows the methods employed. This work is written up for publication in the journal *Social Sciences Research* (detailed below). Within the first nine months of this paper being published, it was downloaded from the Journal's website 1520 times, thus showing the interest in the study design.



(figures above were only provided by the Journal for the timeframe December 2013 – September 2014).

1. 2011-2012

After the background preparation work above, the year 2011-2012 was spent designing the survey, creating the 10 films that sit within it and constructing the recruitment strategy. The films are used as a medium to describe the various ethical issues raised by genomics and are used to offer background information that research participants need in order to answer the questions. The films took 5 months to create from start to finish and involved multiple iterations to get the right balance of information, graphics, filmed footage and voice over. I wrote the scripts for the films and co-directed the filming with Neonotter (professional film making company). I wanted to ensure that the films could be used out of context of the survey and would be appropriate for teaching about genomics and could be applied to ethical considerations about sequencing in both a research and clinical setting. A testimony to the versatility of the films and survey has been demonstrated as they are being used in various teaching settings around the world, for example:

- (i) Prof Greg Fowler, founder of Geneforums, uses the films and survey as part of his Public Health Genomics curriculum for doctoral nursing students at Portland State University, USA. He has also used the survey to stimulate debate in a peer reviewed panel discussion at a US Public Health conference in Oregon. The panel session was called: Public

Health and Genomic Medicine: How do get from here to there? and was delivered at the 2014 Oregon Public Health Association Annual Meeting and Conference, the survey was delivered to delegates as part of their pre-conference preparation for participation in the panel.

- (ii) Dr Natasha Anwar, Associate Professor from Department of Biological Sciences, Forman Christian College in Lahore, Pakistan has used the films in medical student teaching about genomics.
- (iii) Dr Clara Gaff, Program lead of Melbourne Genomics Health Alliance and Prof Sylvia Metcalfe from Melbourne Children's Hospital, Australia, use the survey and films to teach undergraduate biomedical students and postgraduate genetic counselling students.
- (iv) Prof Judith Goodship, Institute of Human Genetics, University of Newcastle uses the survey and films in teaching medical students and also A-level students about genetics.
- (v) Mrs Nicola Wilberforce, biology teacher and author of the Salters' Nuffield Advanced Biology online resources, uses the survey and films as a teaching tool for A-level Biology students and teachers to explore the complex ethics surrounding genomic studies.
- (vi) Dr Linda Battistuzzi, biology lecturer from Dept. of Internal Medicine, Genoa University, Italy uses the survey and films to explore ethics with her biology undergraduate students.

The survey (and films) have been adapted and translated by four independent research groups around the world to enable data gathering with different populations:

- (i) Danish: Professor Ole Mors, Department of Clinical Medicine, Aarhus University and Aarhus University Hospital, Risskov
- (ii) Urdu: Dr Natasha Anwar, Associate Professor from Department of Biological Sciences, Forman Christian College in Lahore, Pakistan
- (iii) Spanish: Prof Allesandra Carnevale, Instituto Nacional de Medicina Genomica, Mexico City, Mexico. This work was presented at the European Society of Human Genetics Conference in Milan 2014.

I also consulted experts in social media and recruitment into social sciences research from University of Cambridge as well as from an external Social Media consultancy in order to create the most appropriate participant recruitment strategy. I designed the website that would hold the survey and describe the background to the DDD ethics research (www.genomethics.org) and this was translated and hosted by the WTSI web team. Aside from the survey design, 2011-2012 was spent introducing the project in a public and professional setting via numerous peer reviewed and invited presentations. See final outputs for details.

Video Outputs (10 x varying lengths)

(images are video screen captures. All videos can be found at www.GenomEthics.org under "DDD" study and appear at the portion of the survey indicated by the captions under each)



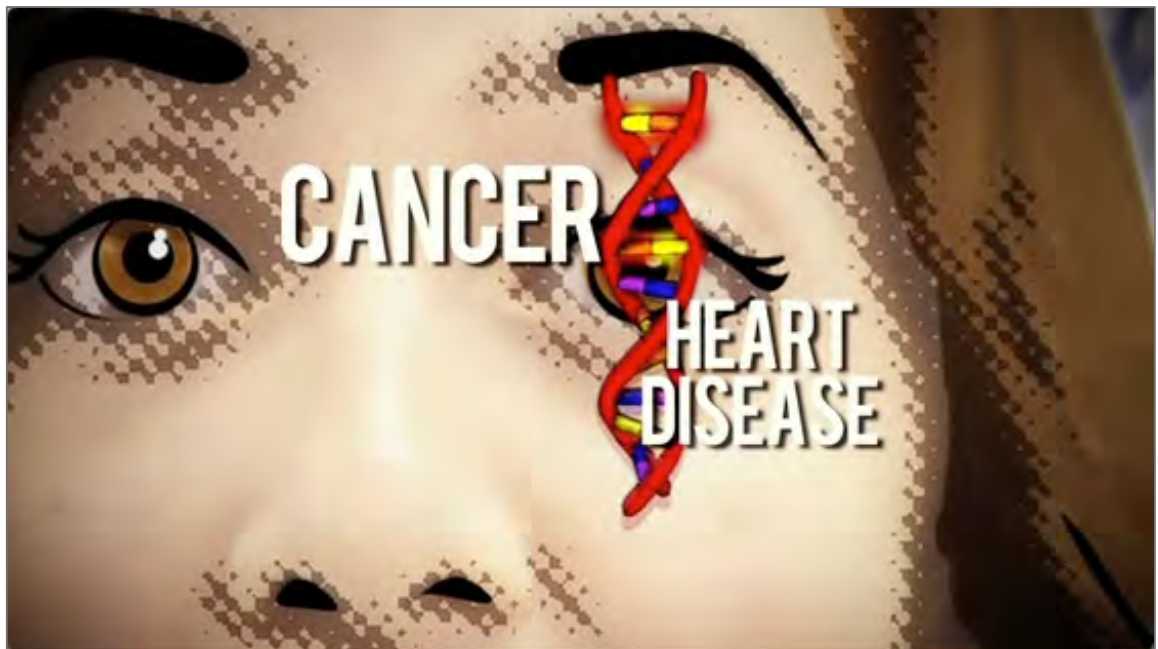
0.) Introduction to Survey



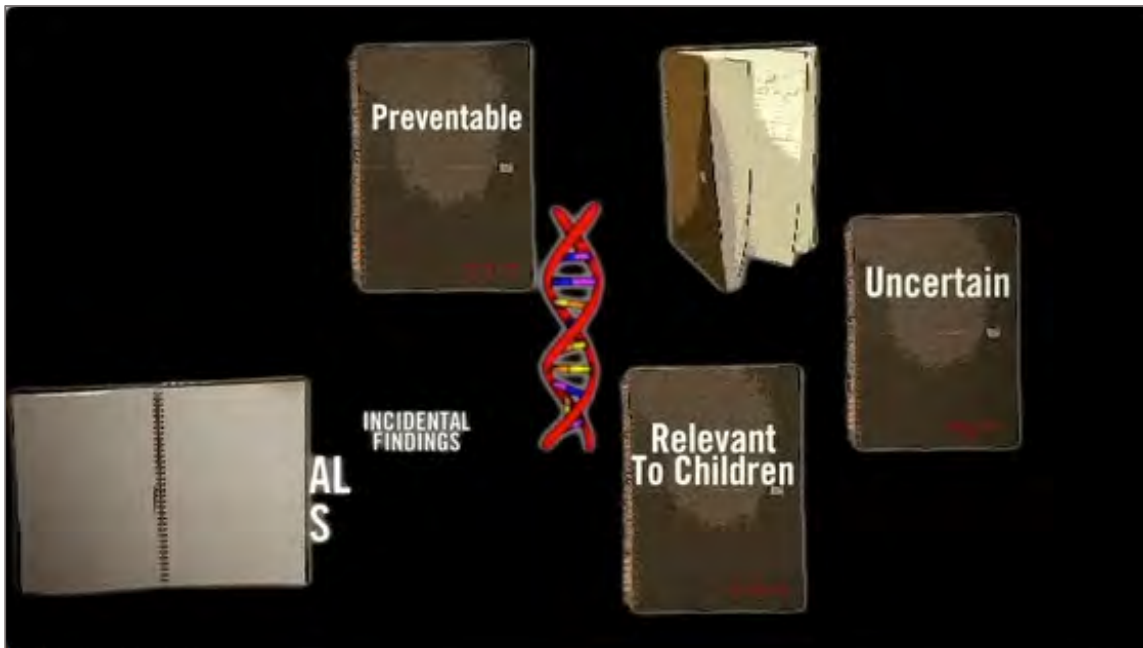
1.) "Questions about you"



2.) "Sharing of Pertinent Findings"



3.) "Sharing of Incidental Findings"



4.) "Categorizing of Incidental Findings"



5.) "Relations with Risk"



6.) "Raw Data"



7.) "Duty of Genomic Researchers"



8.) "Filter of Genomic Information"



9.) "Consent for Genomic Research"

As part of my role as a social scientist researching ethical issues surrounding sequencing technologies I put myself forward for contribution to various external activities both nationally and internationally. This gave me an opportunity to discuss and debate the work I was doing for the DDD Project, as well as explore practical and ethical issues of relevance to genetic counselling practice:

- (i) invited steering group member for NIHR doctoral fellow, Gillian Crawford's PhD project at Southampton University and contributed to discussion about consent and sharing incidental findings;
- (ii) invited member of the Wellcome Trust's Health Related Findings mapping exercise and contributed to their policy creation on sharing incidental findings in research studies across the UK;
- (iii) invited to present to the Human Genetics Commission (before it was disbanded) on the ethical impact of sequencing technologies;
- (iv) invited to be an associate research fellow at the Public Health Genomics Foundation and have participated in their Realising Genomics think tank piece of work exploring the implementation of sequencing in clinic;
- (v) invited 'ethics advisor' to the EBI to explore why a grant had been turned down because the ethical issues had not been addressed, offered advice for a future application;
- (vi) member of the Genome Campus 'Society and Personal Genomics' (SPG) project and as part of this structured the campus genotyping project and wrote up the proposal for consideration by the Board of Management, co-wrote up the REC application, also defended the project in front of the REC. Also co-ordinated and hosted the Social Scientist in Residence sabbatical position for Prof Barb Biesecker and Prof Les Biesecker to visit the Genome Campus for 2 weeks as part of SPG activities;
- (vii) invited member of the Human Materials and Data Management Committee HMDMC Special committee to review applications for work on campus that have an interesting ethical dimension or that have not obtained conventional ethical approval (have worked on 20 proposals to date);
- (viii) invited chair of the Transnational Alliance of Genetic Counsellors in Montreal as part of the International Congress of Human Genetics and American Society of Human Genetics conference;
- (ix) Completed 6 year term (finished June 2011) as vice-chair of the Genetic Counsellor Registration Board UK + ROI - overseeing competency to practice and registration of genetic counsellors in the UK;
- (x) Completed 6 year term as Chair of the Overseas Registration of Genetic Counsellors Working Group (finished June 2011) ;
- (xi) invited co-chair of the International Genetic Counsellor Credentialing Committee, position involved chairing a meeting at the American Society of Human Genetics on the Registration and Certification presidents from UK, USA, Canada, Japan, S. Africa, Australasia;

- (xii) Awarded a highly competitive residency fellowship to spend 1 month at the Brocher Foundation, Geneva (August 2011). Spent time editing a book: 'Getting the message: communication in genetics' published by Oxford University Press.

2. 2012-2013

In January 2012, together with the media team at the Wellcome Trust Sanger Institute, I wrote a press release that would be used as a basis to invite members of the news, radio and written press to report on the ethics study. The aim of this was to promote the online survey and enhance recruitment into the research. I was interviewed about the study for Channel 4 news, BBC Look East news, BBC Radio Cambridgeshire breakfast show and Naked Scientists show, Radio 4 Material World. The focus of this media work was to talk about the GenomEthics study and invite participation in the online survey.

From February 2012 - January 2013 I was on maternity leave. While I was off I arranged for a poster on the ethics study to be presented at three conferences (see outputs for details), I also finished off the necessary proofing required to get the book, with Oxford University Press, into press. I also promoted the online survey where possible so that participants could be recruited into the study in my absence.

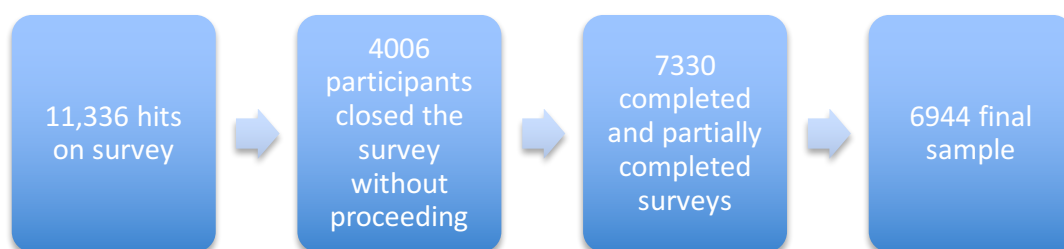
3. 2013-2014

Throughout 2013 after returning from maternity leave I spent the first 5 months doing a social media push to encourage recruitment into the survey. This involved creating a blog where I could discuss current issues in genomics (www.genomethicsblog.org), within the blog page was a large hyperlinked image of the GenomEthics survey together with an invitation to participate in the survey. I also created a Twitter account (@genomethics), a Facebook page (<https://www.facebook.com/Genomethics>), a LinkedIn account (<https://www.linkedin.com/pub/anna-middleton/64/17/895>) and my own personal website (www.annamiddleton.info). The aim of all of these social media outlets was to create an environment where I could actively discuss the survey and related issues and use this activity to increase participation in the study. I posted messages on LinkedIn, Facebook, Twitter and the blog and I also wrote blog posts for the Wellcome Trust, the charity Swan and the charity Unique. This active online debate and promotion was successful and resulted in an increase of participants from 2000 to 7000, with input from 91 different countries across the world. The social media strategy was novel and (as far as I'm aware) had not been done before in this way within social sciences research, I was invited to present this method of recruitment at the HeLEX department, University of Oxford and also at the Sanger Institute (see outputs for details).

4. 2014-2015

I wrote up a second methods paper (see outputs) on the recruitment strategy, together with an overview of the resultant study sample obtained, which was published in *Journal of Community Genetics*. According to the *Journal of Community Genetics* website, as of June 2014, this was the third most popular article they had published and through sharing via Twitter had been made available to 61,000+ followers.

The recruitment strategy was incredibly successful and resulted in over 11,000 hits on the survey. After the data had been cleaned this left just under 7,000 surveys where over 75% of the questions had been completed. All four different stakeholder groups were represented (members of the public, $n = 4961$; genomic researchers, $n = 607$; genetic health professionals, $n = 533$; other health professionals, $n = 843$).



I worked with statistician Dr Kate Morley from Kings College London to fine-tune the statistical analyses. I then began to present the first set of analysis at various conferences in the UK and overseas (see outputs). I also did more media interviews on the ethical implications of sharing genomic data for TV (Royal Society of Science Festival, Wellcome Trust Sanger Institute public engagement material), radio (BBC World Service, Polish Radio) and interviewed for newspaper articles (Observer, Daily Mail, Telegraph, Irish Times) and this time I was able to present some of our own data from the ethics study.

1. 2015-2016

The focus group arm of the study as designed, patient invitation letters, consent forms and a SOP was developed for recruitment. The actual invitation to participate had to be sent by an NHS health professional and this involved a DDD research nurse or clinician checking patient records to make sure that the address was still current and that family circumstances hadn't changed (e.g. a child died). Unfortunately, due to logistical reasons, this was not straightforward. Thus in the time available, only 1 focus group with DDD parents has been possible. I am available and willing to do more focus groups in the future, if and when this logistical issue resolves. While I was waiting for the focus groups to materialise, I spent time writing up the remaining papers on the project and publicising the

results nationally and internationally through conferences and media/online opportunities.

As an extension of my role as DDD Ethics Researcher I have been invited to contribute nationally and internationally as an expert on ethics/genomics issues: (i) invited to chair the Association of Genetic Nurses and Counsellors working group to create and publish a position paper on opportunistic genomic screening on behalf of British genetic counsellors (see outputs for publication that appeared in the European Journal of Human Genetics and Appendix I); (ii) invited Board moderator for the registration of genetic counsellors in the UK; (iii) Vice-Chair of the Association of Genetic Nurses and Counsellors, representing genetic counsellor interests nationally; (iv) invited plenary speaker at the Human Genetics Society of Australasia, 2013, 2014, to present DDD ethics project and own vision for genetic counselling research; (v) Invited teacher on the Wellcome Trust Advanced Courses for professionals programme: Fundamentals of Genetics Jan 2014, Molecular Pathology and Diagnosis of Cancer, teaching about incidental findings; (vi) Invited with other colleagues from the Sanger Institute to input genomics knowledge into the teaching curriculum for health professionals via the NHS National Genetics and Genomics Education Centre, Birmingham; (vii) filmed a series of short pieces on ethics for the NHS e-learning modules on bioinformatics, created by NOWGEN and the NHS Genetics Education Centre; (viii) invited to be an Expert Adviser to Oxford Desk Reference Clinical Genetics by Firth HV and Hurst JA 2nd edition to be published by OUP 2013. Reviewed the text on Communication. (ix) One of seven invited members of the European Molecular Biology Laboratory's Human Data Committee, established to advise the Director General (Heidelberg) and the Director of the EMBL-EBI (Cambridge) on questions arising in relation to accepting, storing and providing access to data related to human research subjects (2013 - present). (x) Invited reviewer for numerous genetics journals, e.g. PLOS ONE, Journal of Genetic Counselling, American Journal of Medical Genetics, Community Genetics, Clinical Genetics, Twin Research and Human Genetics; (xi) invited by the Public Engagement team at the Wellcome Trust Sanger Institute to help design the 'ethics' component of the Sanger stand at the Royal Society Festival of Science, July 2013. We utilised a multi-media approach - consisting of film with actors asking a series of ethics questions delivered on a large TV screen, a second screen delivering a question that visitors could press buttons to answer and a use of several projectors that beamed down genetic 'traits' as words onto visitor's clothes. Exhibition was designed to run concurrently with a similar exhibition at the Smithsonian Museum in Washington DC.



Cutting the ribbon at the Royal Society Festival of Science 2013 Sanger Institute Stand

In 2015 I was offered a core-funded position setting up social science research for the Wellcome Genome Campus and from 1st October 2016 now run this as Head of Society and Ethics Research, Connecting Science at Wellcome Genome Campus.

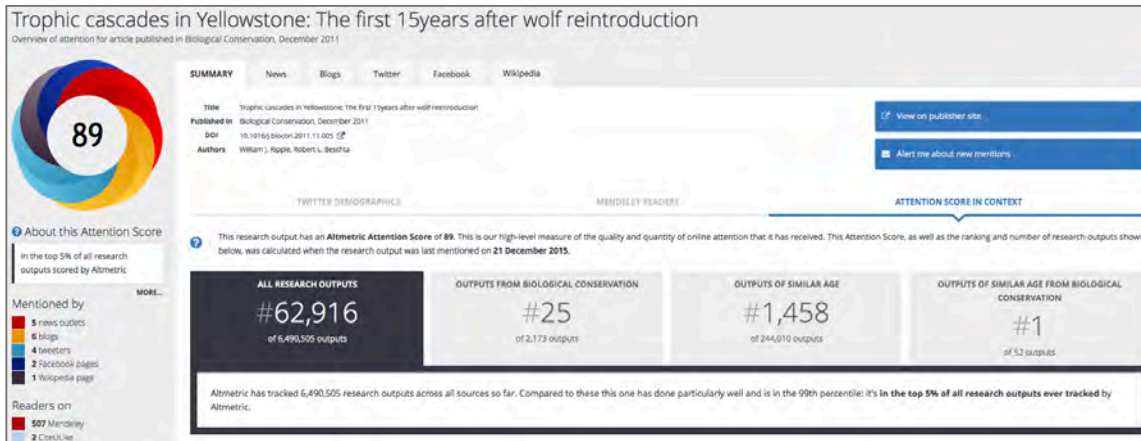
GenomEthics

Impact and Reach of Work

1. Peer Reviewed Journal Articles
2. Mentions of the GenomEthics Study in Select Peer Reviewed Journals and Policy
3. Peer Reviewed Conference Presentations
4. Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study
5. Video, Museum Exhibits and Teaching Materials Based on Outcomes of the GenomEthics Study
6. News Coverage and Media Referring to the GenomEthics Study
7. Blog and Online Journal Articles Referring to the GenomEthics Study
8. Social Media

Interpreting Altmetric Scores and Data

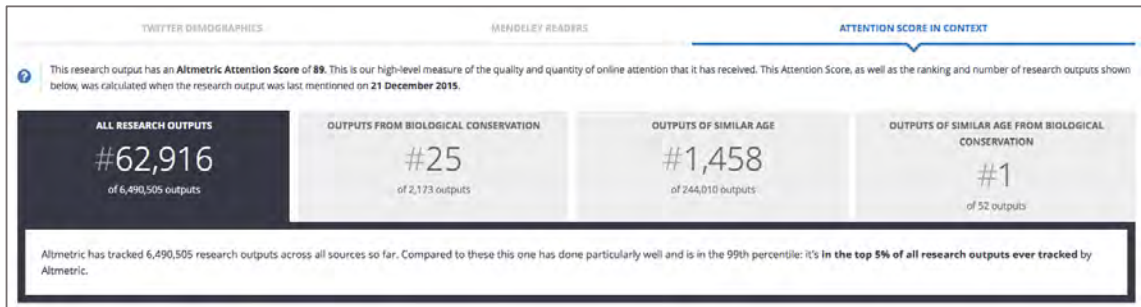
Altmetrics are metrics and qualitative data that are complementary to traditional, citation-based metrics. They can include (but are not limited to) peer reviews on Faculty of 1000, citations on Wikipedia and in public policy documents, discussions on research blogs, mainstream media coverage, bookmarks on reference managers like Mendeley, and mentions on social networks such as Twitter. Altmetric searches for data from a broad range of sources using DOIs and then collates that data into an “Altmetric Attention Score”.



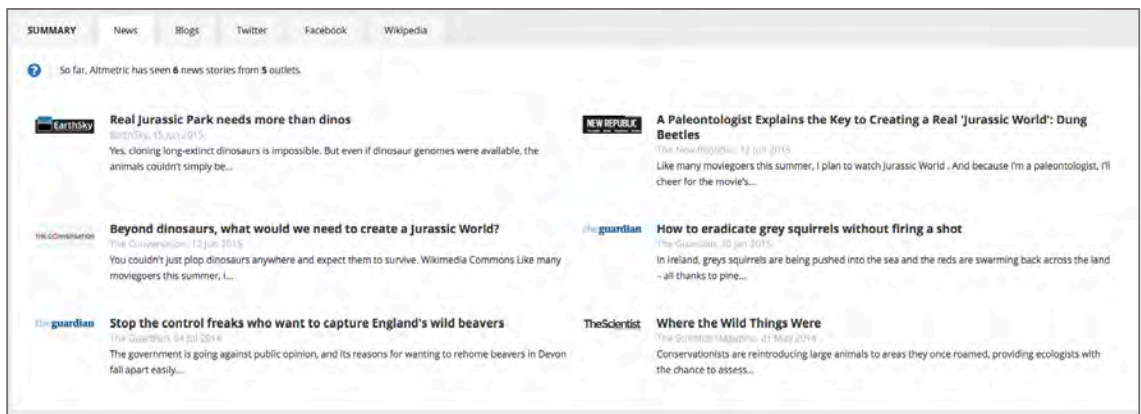
The colours in the doughnut represent various types of media (as indicated below the doughnut), the more colourful the doughnut the more varied types of media that article has reached.

The score, located in the centre of the doughnut, is derived from an automated algorithm, and represents a weighted count of the amount of attention we've picked up for a research output. It is weighted to reflect the relative reach of each type of source (to account for the fact that the average newspaper story is more likely to bring attention to the research output than the average tweet) The default weightings are as follows:

News	8
Blogs	5
Twitter	1
Facebook	0.25
Sina Weibo	1
Wikipedia	3
Policy Documents (per source)	3
Q&A	0.25
F1000/Publons/Pubpeer	1
YouTube	0.25
Reddit/Pinterest	0.25
LinkedIn	0.5



“Attention Score in Context” shows where that particular output ranks in relation to all research outputs, other outputs from the same publication and where it specifically ranks in relation to other outputs of a similar age.



It is possible to drill down to see the specific instances that the output appeared in various media (in the below example, the “News” tab is shown) and then follow the link back to the story at its original source.

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Morley K, Bragin E, Firth HV, Hurles M, Wright CF, Parker M on behalf of the DDD study (2016) Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research. *European Journal Human Genetics* 24(1): p21-29 (*Altmetric data gathered 20 Sept 2016*)

European Journal of Human Genetics - Attitudes of nearly 7000 hea... toward the return of incidental results from sequencing research 20/09/2016, 12:56

Article

European Journal of Human Genetics (2016) **24**, 21–29; doi:10.1038/ejhg.2015.58; published online 29 April 2015


Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research

EJHG Open

Anna Middleton¹, Katherine I Morley^{2,3}, Eugene Bragin¹, Helen V Firth⁴, Matthew E Hurles¹, Caroline F Wright¹ and Michael Parker⁵ on behalf of the DDD study

Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research

Overview of attention for article published in *European Journal of Human Genetics*, April 2015



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SUMMARY News Blogs Twitter Facebook

Title: Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research
Published in: *European Journal of Human Genetics*, April 2015
DOI: 10.1038/ejhg.2015.58
PubMed ID: 25920556
Authors: Anna Middleton, Katherine I Morley, Eugene Bragin, Helen V Firth, Matthew E Hurles, Caroline F Wright
Abstract: Genome-wide sequencing in a research setting has the potential to reveal health-related...

ATTENTION SCORE IN CONTEXT

This research output has an **Altmetric Attention Score** of 193. This is our high-level measure of the quality and quantity of online attention that it has received. This Attention Score, as well as the ranking and number of research outputs shown below, was calculated when the research output was last mentioned on **01 September 2016**.

Category	Score	Context
ALL RESEARCH OUTPUTS	#20,574	of 6,368,878 outputs
OUTPUTS FROM EUROPEAN JOURNAL OF HUMAN GENETICS	#3	of 1,605 outputs
OUTPUTS OF SIMILAR AGE	#1,113	of 168,766 outputs
OUTPUTS OF SIMILAR AGE FROM EUROPEAN JOURNAL OF HUMAN GENETICS	#2	of 81 outputs

Altmetric has tracked 6,368,878 research outputs across all sources so far. Compared to these this one has done particularly well and is in the 99th percentile: it's in the **top 5% of all research outputs ever tracked** by Altmetric.

Mentioned by: 10 news outlets, 1 blog, 166 tweeters, 8 facebook pages

Readers on: 48 Mandelley, 4 CiteULike

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Morley K, Bragin E, Firth HV, Hurler M, Wright CF, Parker M on behalf of the DDD study (2016) Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research. *European Journal Human Genetics* 24(1): p21-29 (continued)

Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research
 Overview of attention for article published in *European Journal of Human Genetics*, April 2015

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So far, Altmetric has seen **10** news stories from **10** outlets.

What Digital Innovation in Health Care Will Look Like
The New York Observer, 02 May 2015
 Digitization of healthcare records has literally paved the way for a far more informed, innovative, and personalized care...

What Digital Innovation in Health Care Will Look Like
Business Insider (New York Observer), 02 May 2015
 Digitization of healthcare records has literally paved the way for a far more informed, innovative, and personalized care...

Volunteering for a genetic study, would you want to know?
Digital Journal, 10 May 2015
 The majority of people who participate in research that involves genetic testing prefer to be told if they have mutations that...

Most people eager to know the secrets of their genetics
Medical News Today, 01 May 2015
 Largest survey of public attitudes shows perceived value of genomic data: A survey of nearly 7000 people has revealed that 98 per...

98 Percent Want to Know if Genetic Data Contains A Serious Preventable Or Treatable Disease
Science 2.0, 29 Apr 2015
 A survey of nearly 7,000 people found that 98 percent want to know if their genetic data contains indicators of a serious...

Attitudes of nearly 7000 health professionals genomic researchers and publics toward the return of incidental results from sequencing research
Sciortone, 29 Apr 2015

Most people eager to know the secrets of their genetics
Medical Xpress, 25 Apr 2015
 A survey of nearly 7000 people has revealed that 98 per cent want to be informed if researchers using their genetic data...

Study Participants Want to Know
The Scientist Magazine, 28 Apr 2015
 Most people who participate in research that involves genetic testing prefer to be told if they have mutations that increase...

Survey Shows Research Participants Want to Know Genomic Testing Information
Genomeweb, 29 Apr 2015

Most people eager to know the secrets of their genetics
EurekaAlert, 29 Apr 2015
 [Wellcome Trust Sanger Institute] 98 percent of people want to be told about indicators of a serious preventable or treatable...

About this Attention Score

- In the top 5% of all research outputs scored by Altmetric
- One of the highest-scoring outputs from this source (#3 of 1,605)
- High Attention Score compared to outputs of the same age (99th percentile)
- High Attention Score compared to outputs of the same age and source (97th percentile)

Mentioned by

- 10 news outlets
- 1 blog
- 166 tweeters
- 8 Facebook pages

Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research
 Overview of attention for article published in *European Journal of Human Genetics*, April 2015

193

So far, Altmetric has seen **174** tweets from **166** users, with an upper bound of **542,587** followers.

Showing items 1-100

Meg Jones (@msthesdijones)
 Some great things in the new issue of @nature EJHG including perceptions of disclosing incidental genetic findings <https://t.co/yXah9lqR1>
 21 Dec 2015

Anne (@anneTHEcaptain)
 People want their DNA sequencing data—all of it—and even more so than medical professionals <https://t.co/rIg5s5xE4Z> <https://t.co/Xj5Eu0e0j>
 19 Jun 2015

Janett Wawrzyniak (@wawrzyniak9)
 Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental... <http://t.co/S1M46uTAk>
 15 Jun 2015

Viswanathan Sathesh (@satheshbhcg)
 People want their DNA sequencing data—all of it—and even more so than medical professionals <https://t.co/rIg5s5xE4Z> <https://t.co/Xj5Eu0e0j>
 12 Jun 2015

Nicole Holm (@nicholasholm)
 People want their DNA sequencing data—all of it—and even more so than medical professionals <https://t.co/rIg5s5xE4Z> <https://t.co/Xj5Eu0e0j>
 11 Jun 2015

Kevin Douglas Moran (@KDMHealthUpdate)
 Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental resul <https://t.co/33Efc94to>
 14 Dec 2015

Somalee (@somaleedata)
 People want their DNA sequencing data—all of it—and even more so than medical professionals <https://t.co/rIg5s5xE4Z> <https://t.co/Xj5Eu0e0j>
 19 Jun 2015

Abhiman Saraswathi (@abhimsans)
 People want their DNA sequencing data—all of it—and even more so than medical professionals <https://t.co/rIg5s5xE4Z> <https://t.co/Xj5Eu0e0j>
 12 Jun 2015

Karen O'Leary (@kmo02)
 People want their DNA sequencing data—all of it—and even more so than medical professionals <https://t.co/rIg5s5xE4Z> <https://t.co/Xj5Eu0e0j>
 11 Jun 2015

Kirk Willmarth (@kirkwillmarth)
 People want their DNA sequencing data—all of it—and even more so than medical professionals <https://t.co/rIg5s5xE4Z> <https://t.co/Xj5Eu0e0j>
 11 Jun 2015

About this Attention Score

- In the top 5% of all research outputs scored by Altmetric
- One of the highest-scoring outputs from this source (#3 of 1,605)
- High Attention Score compared to outputs of the same age (99th percentile)
- High Attention Score compared to outputs of the same age and source (97th percentile)

Mentioned by

- 10 news outlets
- 1 blog
- 166 tweeters
- 8 Facebook pages

Readers on

- 48 Mentions
- 4 CiteULike

What is this page?

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Wright CF, Morley KI, Bragin E, Firth HV, Hurles ME, Parker M on behalf of the DDD study (2015) Potential research participants support the return of raw sequence data. *Journal Medical Genetics* 52(8): p571-574 (*Altmetric data gathered 20 Sept 2016*)

Downloaded from <http://jmg.bmj.com/> on September 20, 2016 - Published by group.bmj.com
JMG Online First, published on May 20, 2015 as 10.1136/jmedgenet-2015-103119 Clinical guidelines

 **OPEN ACCESS**


SHORT REPORT

Potential research participants support the return of raw sequence data

Anna Middleton,¹ Caroline F Wright,¹ Katherine I Morley,^{2,3} Eugene Bragin,¹ Helen V Firth,⁴ Matthew E Hurles,¹ Michael Parker,⁵ on behalf of the DDD study

Potential research participants support the return of raw sequence data.

Overview of attention for article published in *Journal of Medical Genetics*, May 2015



26

About this Attention Score
In the top 5% of all research outputs scored by Altmetric.

Mentioned by
38 tweeters
4 Facebook pages

Readers on
19 Mendeley
2 CiteULike

What is this page?

SUMMARY Twitter Facebook

Title Potential research participants support the return of raw sequence data.
Published in *Journal of Medical Genetics*, May 2015
DOI 10.1136/jmedgenet-2015-103119
PubMed ID 25995218
Authors Anna Middleton, Caroline F Wright, Katherine I Morley, Eugene Bragin, Helen V Firth, Matthew E...
Abstract Health-related results that are discovered in the process of genomic research should only be...

TWITTER DEMOGRAPHICS **MENDELEY READERS** **ATTENTION SCORE IN CONTEXT**

This research output has an **Altmetric Attention Score of 26**. This is our high-level measure of the quality and quantity of online attention that it has received. This Attention Score, as well as the ranking and number of research outputs shown below, was calculated when the research output was last mentioned on **03 August 2015**.

ALL RESEARCH OUTPUTS	OUTPUTS FROM JOURNAL OF MEDICAL GENETICS	OUTPUTS OF SIMILAR AGE	OUTPUTS OF SIMILAR AGE FROM JOURNAL OF MEDICAL GENETICS
#221,797 of 6,248,417 outputs	#19 of 1,372 outputs	#10,647 of 168,388 outputs	#2 of 36 outputs

Altmetric has tracked 6,248,417 research outputs across all sources so far. Compared to these this one has done particularly well and is in the 96th percentile: it's **in the top 5% of all research outputs ever tracked by Altmetric.**

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Hall G, Patch C (2015) Genetic counsellors and Genomic Counselling in the United Kingdom. *Molecular Genetics and Genomic Medicine* 3(2): p79-83 (*Altmetric data gathered 8 Nov 2016*)

Molecular Genetics & Genomic Medicine

Open Access

INVITED COMMENTARY

Genetic counselors and Genomic Counseling in the United Kingdom


Anna Middleton¹, Georgina Hall² & Christine Patch^{3,4}

¹Wellcome Trust Sanger Institute, Cambridge, United Kingdom
²Manchester Centre for Genomic Medicine, Central Manchester University Hospitals NHS Foundation Trust, Manchester Academic Health Sciences Centre (MAHSC), St. Mary's Hospital, Manchester, United Kingdom
³Guys and St Thomas' NHS Foundation Trust, London, United Kingdom
⁴Kings College London, London, United Kingdom

doi: 10.1002/mgg3.123

Genetic counselors and Genomic Counseling in the United Kingdom

Overview of attention for article published in *Molecular Genetics & Genomic Medicine*, December 2014



25

Summary: Title: Genetic counselors and Genomic Counseling in the United Kingdom; Published in: *Molecular Genetics & Genomic Medicine*, December 2014; DOI: 10.1002/mgg3.123; Pubmed ID: 25802877; Authors: Middleton, Anna, Hall, Georgina, Patch, Christine

Twitter Demographics: 37 tweeters, 3 Facebook pages, 1 video uploader

Readers on: 11 Mendeley

Attention Score in Context: This research output has an **Altmetric Attention Score of 25**. This is our high-level measure of the quality and quantity of online attention that it has received. This Attention Score, as well as the ranking and number of research outputs shown below, was calculated when the research output was last mentioned on **07 June 2015**.

Category	Score	Context
All Research Outputs	#248,169	of 6,529,078 outputs
Outputs from Molecular Genetics & Genomic Medicine	#4	of 141 outputs
Outputs of Similar Age	#12,513	of 209,639 outputs
Outputs of Similar Age from Molecular Genetics & Genomic Medicine	#1	of 11 outputs

Altmetric has tracked 6,529,078 research outputs across all sources so far. Compared to these this one has done particularly well and is in the 96th percentile: it's in the **top 5% of all research outputs ever tracked by Altmetric**.

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Parker M, Bragin E, Morley K, Wright C, Firth HV, Hurles M on behalf of the DDD study (2014) No expectation to share incidental findings in genomic research. *Lancet* 385(9975): p1289-1290 (*Altmetric data gathered 20 Sept 2016*)

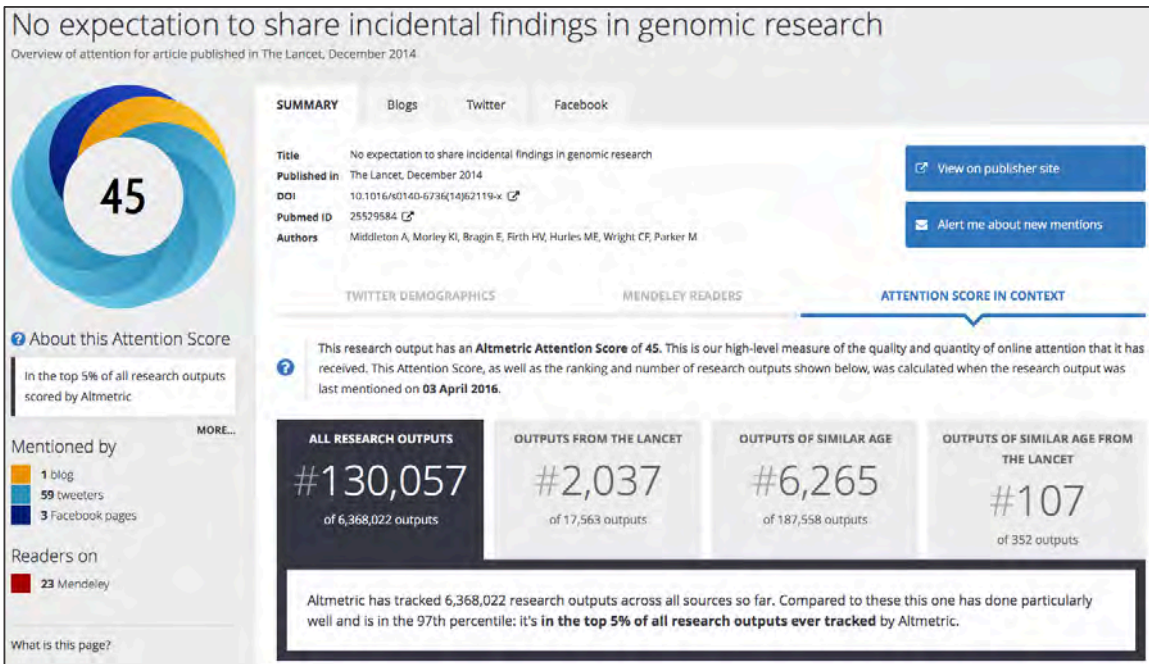


THE LANCET
Volume 385, Issue 9975, 4–10 April 2015, Pages 1289–1290

Correspondence

No expectation to share incidental findings in genomic research

Anna Middleton^a, Katherine I Morley^{b, c}, Eugene Bragin^a, Helen V Firth^d, Matthew E Hurles^a, Caroline F Wright^a, Michael Parker^a, on behalf of the Deciphering Developmental Disorders Study



No expectation to share incidental findings in genomic research
Overview of attention for article published in *The Lancet*, December 2014

45

Summary: No expectation to share incidental findings in genomic research
Published in: *The Lancet*, December 2014
DOI: 10.1016/S0140-6736(14)62119-x
Pubmed ID: 25529584
Authors: Middleton A, Morley KI, Bragin E, Firth HV, Hurles ME, Wright CF, Parker M

Twitter Demographics: 1 blog, 59 tweeters, 3 Facebook pages
Readers on: 23 Mendeley

ATTENTION SCORE IN CONTEXT

This research output has an **Altmetric Attention Score of 45**. This is our high-level measure of the quality and quantity of online attention that it has received. This Attention Score, as well as the ranking and number of research outputs shown below, was calculated when the research output was last mentioned on **03 April 2016**.

ALL RESEARCH OUTPUTS	OUTPUTS FROM THE LANCET	OUTPUTS OF SIMILAR AGE	OUTPUTS OF SIMILAR AGE FROM THE LANCET
#130,057 of 6,368,022 outputs	#2,037 of 17,563 outputs	#6,265 of 187,558 outputs	#107 of 352 outputs

Altmetric has tracked 6,368,022 research outputs across all sources so far. Compared to these this one has done particularly well and is in the 97th percentile: it's in the **top 5% of all research outputs ever tracked** by Altmetric.

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Bragin E, Morley KI, Parker M on behalf of the DDD Study (2014) Online questionnaire development: using film to engage participants and then gather attitudes towards the sharing of genomic data. Social Science Research 44: p211-223
(Altmetric data gathered 20 Sept 2016)

Social Science Research 44 (2014) 211-223

Contents lists available at ScienceDirect

Social Science Research

journal homepage: www.elsevier.com/locate/ssresearch

Online questionnaire development: Using film to engage participants and then gather attitudes towards the sharing of genomic data

A. Middleton ^{a,*}, E. Bragin ^a, K.I. Morley ^{b,c}, M Parker ^d, on behalf of the DDD Study ^a

(Note: The image also shows Elsevier and CrossMark logos.)

Online questionnaire development: Using film to engage participants and then gather attitudes towards the sharing of genomic data.

Overview of attention for article published in Social Science Research, March 2014

13

Summary | Twitter

Title Online questionnaire development: Using film to engage participants and then gather attitudes towards the sharing of genomic data. [View on publisher site](#)

Published in Social Science Research, March 2014

DOI 10.1016/j.ssresearch.2013.12.004 [↗](#)

Pubmed ID 24468445 [↗](#)

Authors Middleton A, Bragin E, Morley KI, Parker M

Abstract How can a researcher engage a participant in a survey, when the subject matter may be perceived as "... [\[show\]](#)

ATTENTION SCORE IN CONTEXT

This research output has an **Altmetric Attention Score of 13**. This is our high-level measure of the quality and quantity of online attention that it has received. This Attention Score, as well as the ranking and number of research outputs shown below, was calculated when the research output was last mentioned on **30 January 2014**.

ALL RESEARCH OUTPUTS	OUTPUTS FROM SOCIAL SCIENCE RESEARCH	OUTPUTS OF SIMILAR AGE	OUTPUTS OF SIMILAR AGE FROM SOCIAL SCIENCE RESEARCH
#361,404 of 4,771,353 outputs	#76 of 431 outputs	#13,045 of 128,206 outputs	#3 of 8 outputs

Altmetric has tracked 4,771,353 research outputs across all sources so far. Compared to these this one has done particularly well and is in the 92nd percentile: it's in the **top 10% of all research outputs ever tracked** by Altmetric.

About this Attention Score

In the top 25% of all research outputs scored by Altmetric

Mentioned by **17** tweeters

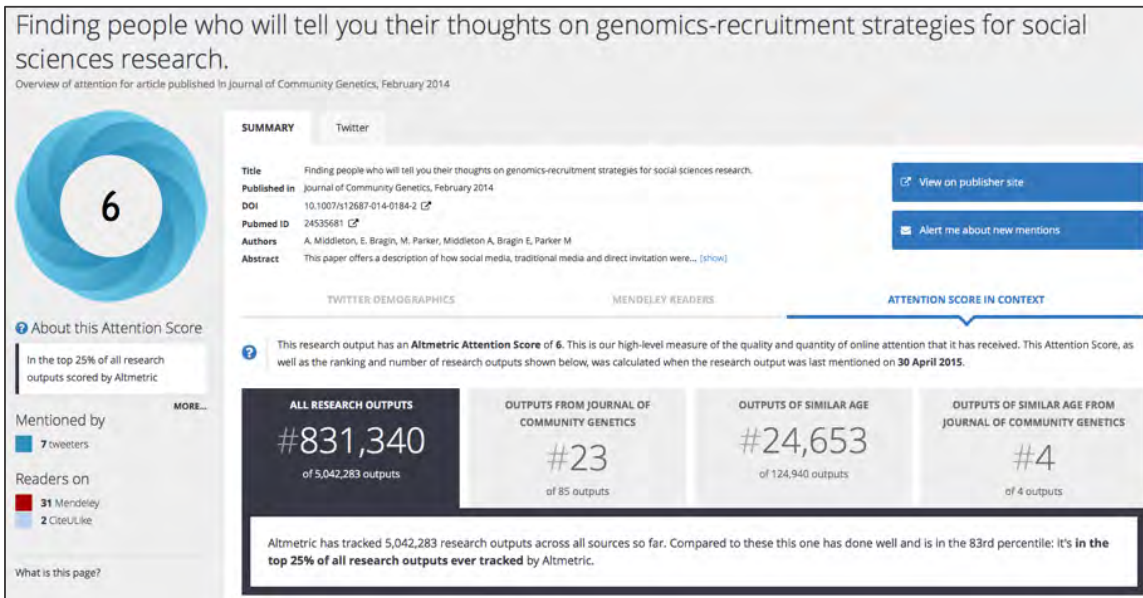
Readers on **39** Mendeley

What is this page?

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Bragin E, Parker M on behalf of the DDD Study (2014)
Finding people who will tell you their thoughts on genomics -
recruitment strategies for social sciences research. J Community
Genetics. 5: p291-302 (*Altmetric data gathered 20 Sept 2016*)



Middleton A, Patch C, Wiggins J, Barnes K, Crawford G, Benjamin C,
Bruce A on behalf of the Association of Genetic Nurses and
Counsellors in the United Kingdom and Ireland (2014) Position
statement on opportunistic genomic screening from the Association of
Genetic Nurses and Counsellors (UK and Ireland)
European Journal Human Genetics 22: p955-956
(*Altmetric data not available*)

Peer Reviewed Journal Articles

(section 1 of 8)

Middleton A, Parker M, Wright CF, Bragin E, Hurles M on behalf of the DDD Study (2013). Empirical Research on the Ethics of Genomic Research. American Journal Medical Genetics Part A 161A: p2099-2101 (*Altmetric data gather 8 Nov 2016*)

The image shows the front cover of a journal article. On the left, a blue box contains the text 'RESEARCH LETTER'. On the right, the journal title 'AMERICAN JOURNAL OF medical genetics' is displayed with a 'PART A' logo. The main title 'Empirical Research on the Ethics of Genomic Research' is centered in a large blue font. Below the title, the authors are listed: 'Anna Middleton,^{1*} Michael Parker,² Caroline F. Wright,¹ Eugene Bragin,¹ Matthew E. Hurles,¹ and on behalf of the DDD Study'.

The image is a screenshot of an Altmetric attention score dashboard. At the top, it displays the title 'Empirical research on the ethics of genomic research' and a sub-header 'Overview of attention for article published in American Journal of Medical Genetics, Part A, January 2013'. A large circular graphic on the left shows an attention score of 3. Below this, there are sections for 'About this Attention Score', 'Mentioned by' (3 tweeters, 1 Facebook page), and 'Readers on' (16 Mendeley, 3 CiteULike). The main content area is divided into tabs for 'SUMMARY', 'Twitter', and 'Facebook'. The 'SUMMARY' tab is active, showing the article's title, publication details, DOI (10.1002/ajmg.a.36067), PubMed ID (23813696), and authors. Below the summary, there are sections for 'TWITTER DEMOGRAPHICS', 'MENDELEY READERS', and 'ATTENTION SCORE IN CONTEXT'. The 'ATTENTION SCORE IN CONTEXT' section features four data points: '#2,168,954 of 6,512,394 outputs' for ALL RESEARCH OUTPUTS; '#648 of 1,914 outputs' for OUTPUTS FROM AMERICAN JOURNAL OF MEDICAL GENETICS, PART A; '#31,289 of 101,207 outputs' for OUTPUTS OF SIMILAR AGE; and '#5 of 22 outputs' for OUTPUTS OF SIMILAR AGE FROM AMERICAN JOURNAL OF MEDICAL GENETICS, PART A. A footer note states: 'Altmetric has tracked 6,512,394 research outputs across all sources so far. This one has received more attention than most of these and is in the 66th percentile.'

Middleton A (2012) Communication about DTC testing: commentary on a 'Family Experience of Personal Genomics'. Journal of Genetic Counseling 21(3): p392-398

Peer Reviewed Journal Articles

(section 1 of 8)

****Main Project Paper (included for comparison)****

Wright, Caroline F et al. on behalf of the DDD study (2014). Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet 385(9975): p1305-1314
(Altmetric data gathered 20 Sept

THE LANCET

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All Content Search Advanced Search

< Previous Article Volume 385, No. 9975, p1305–1314, 4 April 2015 Next Article >

Articles

Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data

Dr Caroline F Wright, PhD, Tomas W Fitzgerald, MS, Wendy D Jones, MBBS, Stephen Clayton, MRes, Jeremy F McRae, PhD, Margriet van Kogelenberg, PhD, Daniel A King, MD, Kirsty Ambridge, BSc, Daniel M Barrett, BSc, Tanya Bayzvetinova, BSc, A Paul Bevan, PhD, Eugene Bragin, MSc, Eleni A Chatzimichali, PhD, Susan Gribble, PhD, Philip Jones, MSc, Netravathi Krishnappa, MSc, Laura E Mason, BSc, Ray Miller, PhD, Katherine I Morley, PhD, Vijaya Parthiban, PhD, Elena Prigmore, PhD, Diana Rajan, MSc, Alejandro Sifrim, PhD, G Jawahar Swaminathan, PhD, Adrian R Tivey, MSc, Anna Middleton, PhD, Prof Michael Parker, PhD, Nigel P Carter, PhD, Jeffrey C Barrett, PhD, Matthew E Hurles, PhD, Prof David R FitzPatrick, DM, Helen V Firth, DM, on behalf of the DDD study¹

Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data

Overview of attention for article published in The Lancet, December 2014

145

Summary News Blogs Twitter Facebook Research highlights

Title Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data

Published in The Lancet, December 2014

DOI 10.1016/S0140-6736(14)61705-0

Pubmed ID 25529582

Authors Wright CF, Fitzgerald TW, Jones WD, Clayton S, McRae JF, van Kogelenberg M, King DA, Ambridge K... (show)

Abstract Human genome sequencing has transformed our understanding of genomic variation and its relevance... (show)

View on publisher site

Alert me about new mentions

TWITTER DEMOGRAPHICS MENDELEY READERS ATTENTION SCORE IN CONTEXT

This research output has an **Altmetric Attention Score** of 145. This is our high-level measure of the quality and quantity of online attention that it has received. This Attention Score, as well as the ranking and number of research outputs shown below, was calculated when the research output was last mentioned on **29 August 2016**.

ALL RESEARCH OUTPUTS	OUTPUTS FROM THE LANCET	OUTPUTS OF SIMILAR AGE	OUTPUTS OF SIMILAR AGE FROM THE LANCET
#30,744	#594	#1,538	#33
of 6,342,725 outputs	of 17,524 outputs	of 186,997 outputs	of 352 outputs

Altmetric has tracked 6,342,725 research outputs across all sources so far. Compared to these this one has done particularly well and is in the 99th percentile: it's in the **top 5% of all research outputs ever tracked by Altmetric**.

About this Attention Score

In the top 5% of all research outputs scored by Altmetric

Mentioned by

- 5 news outlets
- 3 blogs
- 127 tweeters
- 4 Facebook pages
- 1 research highlight platform

Readers on

- 196 Mendeley
- 2 CiteULike

GENOMETHICS

DDD/Ethics Project

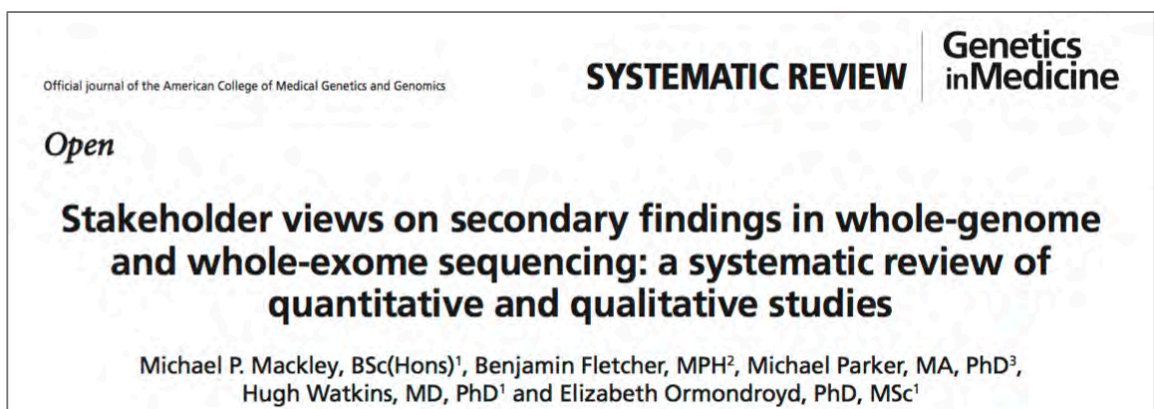
Mentions of the GenomEthics Study in Selected Peer Reviewed Journals and Policy

(section 2 of 8)

Mackley M (2016) Stakeholder views on secondary findings in whole-genome and whole-exome sequencing: a systematic review of quantitative and qualitative studies. *Genetics in Medicine* [online] Volume 1478-6990, p1-11 [Accessed on 16 Nov. 2016] Available at <http://www.nature.com/gim/journal/vaop/ncurrent/full/gim2016109a.html>



The screenshot shows a blog post from the Harvard Law Petrie-Flom Center. The header includes the center's logo and the title "Bill of Health: Examining the intersection of law and health care, biotech & bioethics". The post is dated September 1, 2016, by kmurphy. The main title of the post is "Considering stakeholders in policy around secondary findings in genomics" by Michael Mackley. Navigation links for "Previous" and "Next" are visible.



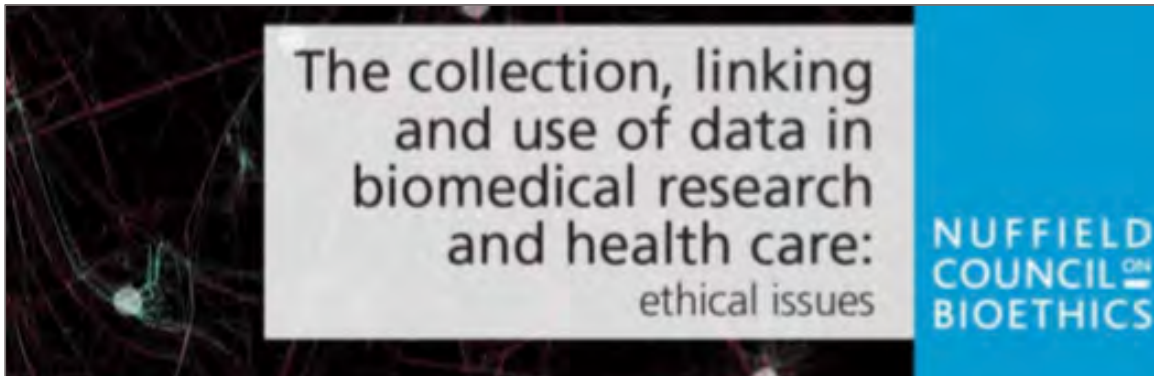
The screenshot shows the cover of a journal article. It is the official journal of the American College of Medical Genetics and Genomics, titled "Genetics in Medicine". The article is a "SYSTEMATIC REVIEW" and is marked as "Open". The title is "Stakeholder views on secondary findings in whole-genome and whole-exome sequencing: a systematic review of quantitative and qualitative studies". The authors listed are Michael P. Mackley, Benjamin Fletcher, Michael Parker, Hugh Watkins, and Elizabeth Ormondroyd.

Mentions of the GenomEthics Study in Selected Peer Reviewed Journals and Policy

(section 2 of 8)

The Nuffield Council on Bioethics (2015). The collection, linking and use of data in biomedical research and health care: ethical issues

- Spoke at report launch during session called "Data initiatives in biomedical research"
- Wrote blog entry to coincide with launch



Session 2: Data initiatives in biomedical research

Speaker: **Susan Wallace**, member of the Working Party and Lecturer of Population and Public Health Sciences, Department of Health Sciences, University of Leicester

Responders to comment on the report:

Anna Middleton, Senior Staff Scientist, Wellcome Trust Sanger Institute

The screenshot shows a blog post on the Nuffield Council on Bioethics website. The page has a white background with a blue header. The header includes the Nuffield Council on Bioethics logo on the left and navigation links for 'About', 'News', and 'Blog' on the right. Below the header, there are three tabs: 'Current work', 'Previous work', and 'Future work'. The main content area features the title 'Exploring ethical issues in biology and medicine' and a breadcrumb trail 'Home > Blog'. The main heading of the post is '"If you know it, I'd like to know it too" – involving participants in genomics research'. Below the heading, there is a 'Back' link and a 'leave a comment' button. The post details include: 'Posted on March 2nd 2015 by Guest', 'Posted in Biological and health data.', and 'Tagged with: bioinformation, clinical trials, DNA.'. A small profile picture of Dr Anna Middleton is shown next to her name and the text 'Guest post'. At the bottom of the post, there is a summary: 'Guest post by Dr Anna Middleton, from the Wellcome Trust Sanger Institute, who spoke at the launch of the Council's report on biological and health care data.'

GENOMETHICS

DDD/Ethics Project

29

Mentions of the GenomEthics Study in Selected Peer Reviewed Journals and Policy


(section 2 of 8)

Contacted by Deloitte to consult on their forthcoming Office for Life Sciences policy report on the Genomics market across Europe, an expansion on their already published study on the Genomics market in the UK (image below), July 2016



MacArthur, D (2012). Challenges in Clinical Genomics. *Genome Medicine* 4(5), p.45

MacArthur *Genome Medicine* 2012, 4:43
<http://genomemedicine.com/content/4/5/43>



MEETING REPORT

Challenges in clinical genomics

Daniel G MacArthur^{1,2*}

Abstract
A report on the Genomic Disorders 2012: Genomics of Rare Diseases meeting, Hinxton, UK, 21-23 March 2012.

Genomics enters the clinic
The central theme of this year's meeting was very clear: genomic technologies are now entering clinical laboratories across the world on a scale that would have seemed unimaginable even a year ago.
The technology that has made the greatest impact in

generate results that flow (via a process of validation) back to clinicians and patients. The task of assembling systems for translating genomic data into clinical interpretation was thus a key topic of discussion.
This meeting provided an eloquent counterpoint to media reports on the 'failure' of the Human Genome Project: medicine is clearly being transformed by genomics, with rare diseases at the vanguard of that transformation. There has never been a more exciting time to be working in the field.

The interpretation challenge

Peer Reviewed Conference Presentations

(section 3 of 8)

Middleton A on behalf of the DDD project (2016) Engaging 7,000 people about the return of results from sequencing research. International Congress of Human Genetics. CIS18-4. Kyoto, Japan 5th April. Invited Plenary Spoken presentation.



Middleton A, Wright C, Firth H, Hurler M, Parker M on behalf of the DDD study (2015) Attitudes towards returning data to participants in sequencing research. C14.5. European Society Human Genetics, Glasgow 8th June. Spoken presentation.



Peer Reviewed Conference Presentations

(section 3 of 8)

Middleton A (2014) Introduction to the Great Debate: Opportunistic screening. British Society Genetic Medicine annual conference, Liverpool 22-24th September.

Middleton A (2014) Young people and sequencing technologies: confusion and clarity. British Society Genetic Medicine annual conference, Liverpool, 22-24th September. Invited symposia speaker.

Middleton A (2014) What's the fuss about incidental findings? Opportunistic screening and international attitudes. Human Genetics Society of Australasia annual conference, Adelaide, Australia, 4th August. Invited International Plenary speaker.



Middleton A, Parker M, Bragin E, Wright C, Firth H, Hurles M on behalf of the DDD Study (2014) International views on sharing incidental findings from whole genome research. European Society Human Genetics conference. Milan, Italy, 31 May - 3 June. Spoken presentation at joint ESHG/EMPAG session rather than smaller group session.



GENOMETHICS
DDD/Ethics Project

Peer Reviewed Conference Presentations

(section 3 of 8)

Middleton A, Parker M, Bragin E, Wright C, Firth H, Hurles M on behalf of the DDD Study (2014) International views on sharing incidental findings from whole genome research. Genomic Disorders 2014 - the genomics of rare diseases. Cambridge, UK, 5-7 March. Spoken presentation.

Middleton A, Parker M, Bragin E, Wright C, Firth H, Hurles M on behalf of the DDD Study (2013) International views on sharing incidental findings from whole genome research. 63rd American Society of Human Genetics Annual Meeting in Boston, MA October 22-26. Spoken presentation.



Middleton A, Parker M, Bragin E, Wright C, Firth H, Hurles M on behalf of the DDD Study (2013) Sharing incidental findings from whole genome research. British Society Genetic Medicine Annual Conference. Liverpool, UK 16-18 September. Spoken presentation.

Middleton A, Parker M, Bragin E, Wright CF, Bevan AP, Firth H, Hurles M on behalf of the DDD study. (2013) Ethical implications of sharing incidental findings: preliminary findings from an International study. Genomic Disorders Conference, Homerton College, Cambridge 10th-12th April. Poster presentation.

Middleton A, Parker M, Wright C, Firth H, Carter N, Hurles M on behalf of the DDD team (2012) Sharing genomic research data: launch of new study. Association of Genetic Nurses and Counsellors annual conference. Cambridge. April 17th. Poster presentation.

Middleton A, Parker M, Bragin E, Wright CF, Firth H, Carter N, Hurles M on behalf of the DDD team (2012) Sharing data from whole genome studies: empirical study of ethical implications. European Psychosocial Aspects of Genetics Conference. Nürnberg, Germany. June 23 - 26. European Journal Human

Peer Reviewed Conference Presentations

(section 3 of 8)

Genetics 20(1). EP08.03. Poster presentation.

Middleton A, Parker M, Bragin E, Wright CF, Morley K, Bevan AP, Firth H, Carter N, Hurles M on behalf of the DDD team (2012) Sharing genomic research data: launch of an international study. British Society of Human Genetics Conference, Warwick. 17-19 September. Poster presentation.

Middleton A, Parker M, Bragin E, Wright C, Morley K, Bevan AP, Firth H, Carter N, Hurles M on behalf of the DDD Study (2012) Sharing data from whole genome studies: empirical study of ethical implications. The Genomics of Rare Diseases, Cambridge, 21-24 March. Poster presentation.

Middleton A, Parker M, Firth H, Carter N (2011) Exploring the ethics of incidental findings from whole genome studies: understanding what research participants, genomic researchers and genetics professionals want. Abstract 1358T. International Congress of Human Genetics/61st Annual Meeting of the American Society of Human Genetics, Montreal, 11-15 October. Poster presentation.

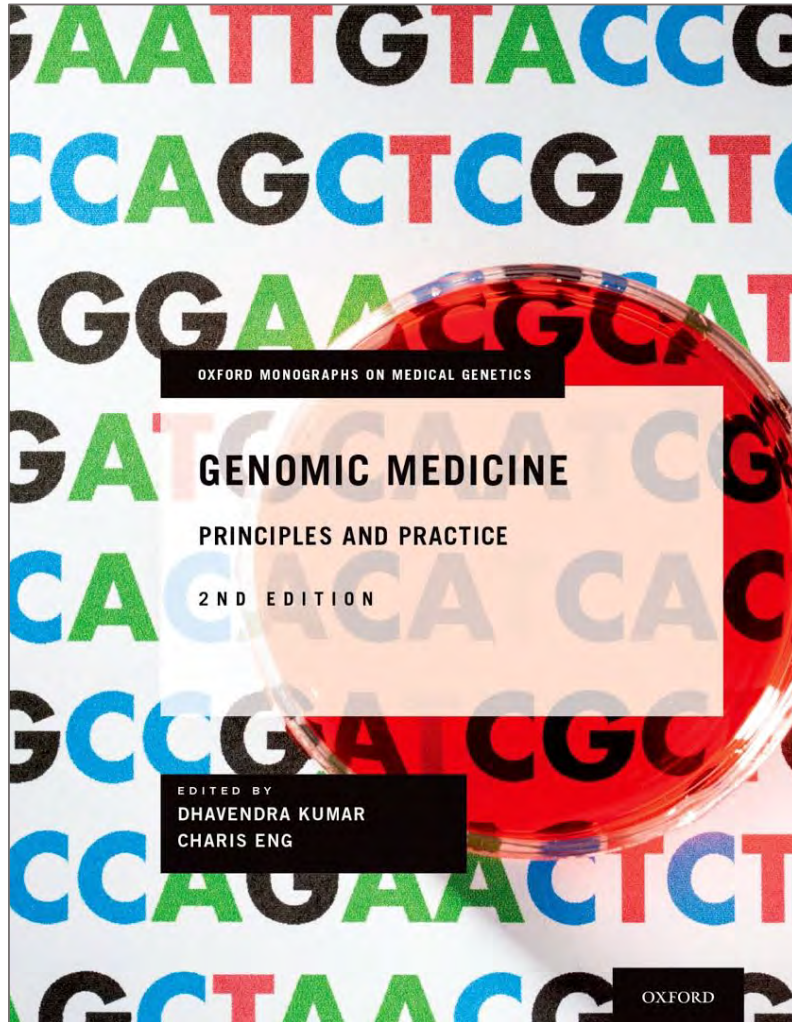
Middleton A, Borthwick G (2011) The central role of genetic counsellors across the UK in the DDD Project. Association of Genetic Nurses and Counsellors Annual Conference, Belfast, 7-9 April. Spoken presentation

Middleton A, Parker M, Firth H, Carter N (2011) Genethics: what to do with incidental findings from whole genome studies? Association of Genetic Nurses and Counsellors Annual Conference, Belfast 7-9 April. Spoken presentation.

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Wright C, **Middleton A**, Parker M (2015) Ethical, legal and social issues in genomics. In: Genomic Medicine: Principles and Practice. Editors: Dhavendra Kumar and Charis Eng. 2nd Edition. New York: Oxford University Press, p250-258



Middleton A (2016) Ethics and Genomics. Teaching on the Wellcome Trust Advanced Course for professionals: Molecular Pathology and Diagnosis of Cancer, Genome Campus, Hinxton, Cambridge, 14th November

Middleton A (2016) Socialising the Genome. Invited Ann McPherson Memorial Lecture. Green Templeton College, 10th October, Oxford

Middleton A (2016) Prioritising Participation: Your Genome, Your Research Agenda. Introduction to Participant GeCIP for Social Science and Ethics. Wellcome Trust, 29th September, London

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2016) Genome Data Privacy. Invited panel member for public event by the British Science Association, supported by Genomics England. Wellcome Collection, 21 September, London

Middleton A (2016) DNA and Big Data. Invited panel member and presenter on Personalised Medicine: The Promise, the Hype and the Pitfalls. University of Oxford. 9th September, Oxford.

Middleton A (2016) Tutor on Public Engagement Masterclass, Conference Centre, Wellcome Genome Campus, Cambridge, 20-22 July

Middleton A (2016) Gathering attitudes from the public towards data sharing: survey and films. CHIPME meeting, 21st May, Barcelona, Spain

Middleton A (2016) Teaching technology and genomics in a rapidly changing environment in the UK. Global trends in genetic counsellor education. Transnational Alliance of Genetic Counselling (TAGC) Fourth International Meeting. The Global State of Genetic Counselling, 20th May, Barcelona, Spain

Middleton A (2016) Invited chair and organiser of the credentialing session for Transnational Alliance of Genetic Counselling (TAGC) Fourth International Meeting. The Global State of Genetic Counselling, 19th May, Barcelona, Spain

Middleton A (2016) Teaching on 2nd year MSc Genetic Counselling course, Cardiff University, 3 hours: Ethics of Genomics and Working with Deaf Clients. 6th May, Cardiff.

Middleton A (2015) If you know it, I'd like to know it too. Article for the Newsletter of the British Society for Genetic Medicine. Issue 53, October 2015

Middleton A (2015) Genomics and social science. Invited seminar to the Genomics and Society group, Kings College London, 1st December

Middleton A (2015) Ethics and Genomics. Teaching on the Wellcome Trust Advanced Course for professionals: Molecular Pathology and Diagnosis of Cancer, Genome Campus, Hinxton, Cambridge, 24th November

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2015) Practical ethics cases. Teaching on Introduction to genomics module of the MSt Genomic Medicine. University of Cambridge, 16th October.

Middleton A (2015) Return of results in participant-centred genomic research. Invited presentation at the Brocher Foundation. Ethical aspects of participant-centred research initiatives. HC Howard, E Vayena, P Borry. Geneva 1-2 October.

Middleton A (2015) Community engagement. Invited presentation for the Global Alliance for Genomics and Health at the European Society Human Genetics conference, Glasgow, 7th June.

Middleton A (2015) Ethics and Social Science. Invited presentation for the DDD Collaborators meeting at the European Society Human Genetics conference, Glasgow, 5th June

Middleton A (2015) Ethics and Technologies. Teaching to campus PhD students, Wellcome Trust Sanger Institute, Cambridge. 22 May.

Middleton A (2015) Ethics and Genomics. Teaching on MSc Genetic Counselling course. Cardiff University. 8th May

Middleton A (2015) Engagement of Research Participants: incidental findings. Global Alliance for Genomics and Health UK Meeting Wellcome Trust, London 24 April 2015

Middleton A (2015) Response to the Nuffield Council on Bioethics report on 'The collection, linking and use of data in biomedical research and health care: ethical issues'. Commentary on the Data Initiatives in Biomedical Research. Westminster, 3rd February

Middleton A (2015) Ethics and Society considerations in the DDD project. Presentation delivered to the Rare Diseases Policy team and Genomics Policy team from the Department of Health. Hinxton Hall, Cambridge. 19th January

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton (2014) Deciphering Developmental Disorders project. Past, Present, Future: The Ethics and Governance of Big Biobanks conference. UK Biobank Ethics and Governance Council. Wellcome Trust, London, 3-5 November

Past, Present, Future:
the Ethics and Governance
of Big Biobanks conference

UK Biobank
Ethics and
Governance
Council

3-5 November 2014
Wellcome Trust, 215 Euston Road, London NW1 2BE



**Monitor
Advise
Safeguard
Report**

14:30 – 16:00	Feedback	<i>Chair and respondent: Dr Jasper Bovenberg</i>
	Wellcome Trust and Medical Research Council Framework on feedback	<i>Ms Katherine Littler</i>
	Framingham Heart Study	<i>Dr Dan Levy</i>
	Deciphering Developmental Disorders project	<i>Dr Anna Middleton</i>
	UK Biobank's policy and the role of the EGC	<i>Dr Sheelagh McGuinness</i>



GENOMETHICS

DDD/Ethics Project

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2014) Attitudes of young people to receiving data from sequencing technologies. RSM Joint meeting RCPCH and RSM Paediatric & Child Health Section and in association with the Clinical Genetics Society. Genomics of paediatric disease. London, 21 October

Middleton A (2014) What's the fuss about incidental findings? Opportunistic screening and international attitudes. Melbourne Genomics Health Alliance and Functional Genomics Series at the Murdoch Children's Research Institute, Melbourne, Australia, 8th August. Invited International Plenary speaker.



Middleton A (2014) What is 'personal genomics'? teaching on the Societal Issues and Personal Genomics course for Biological Sciences third year degree students at University of Melbourne, Melbourne, 7th August

Middleton A (2014) DDD Ethics Study: overview of achievements and future plans. DDD Collaborators Meeting. Cambridge, 23 May

Middleton A (2014) DDD project: Molecular study overview and Ethics overview. Delivery to Peter Goodhand, Executive Director, Global Alliance; Mark Bale, Deputy Head of Health Science and Bioethics, Department of Health; Michael Dunn, Head of Genetic and Molecular Services, Wellcome Trust. Cambridge, 23 May

Middleton A (2014) Incidental findings from sequencing studies: who wants to know what? ethics and attitudes. Association of Clinical Genetic Science annual conference. Birmingham, 29 April

Middleton A (2014) Social media and research. Post-doctoral conference at the Wellcome Trust Sanger Institute. Cambridge, 10 April

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

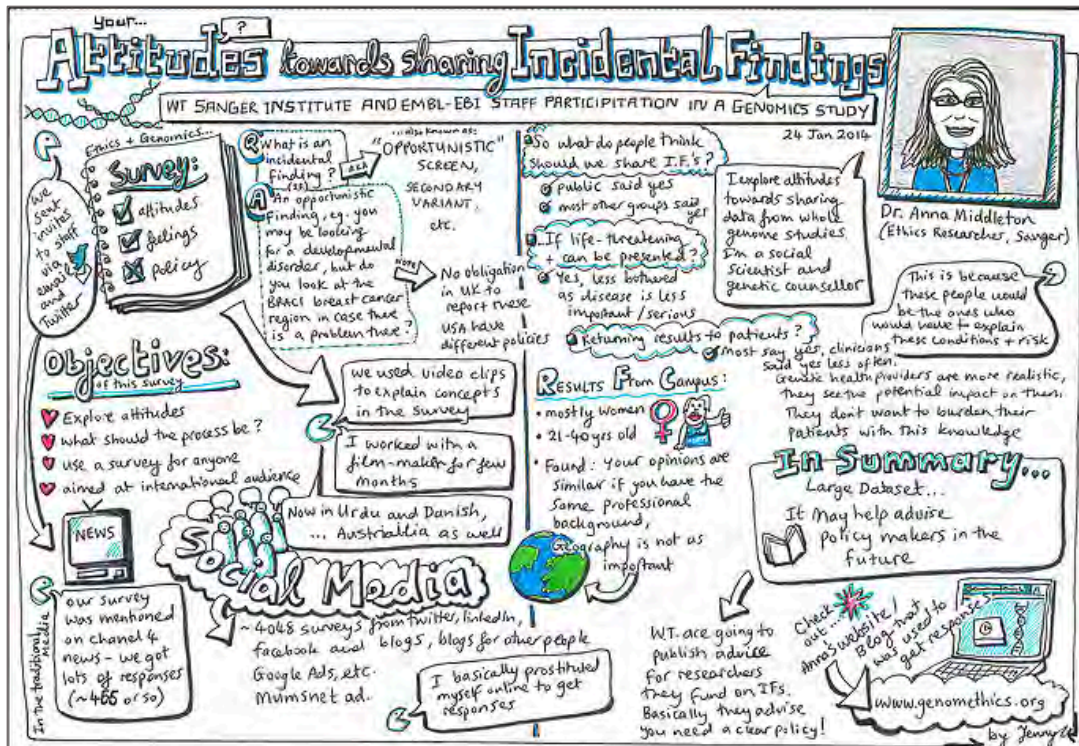
Middleton A (2014) panel discussion together with Sir Mike Stratton and Baroness Helena Kennedy on medical genetics. Names Not Numbers festival of ideas, Aldeborough, Suffolk, 23-25 March



Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2014) Sanger and EBI staff participation in the Genomethics study, your attitudes towards sharing incidental findings from genome research. Wellcome Trust Sanger Institute, Cambridge. 24th January



(Sketch note created by presentation participant Dr. Jennifer Cham to illustrate what was covered)

Middleton A (2014) Genomics, ethics and what people want to know. University of Cambridge. Public Policy. 23rd January

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2014) It's in your genes: what would you want to know? Cafe Scientifique. Bishops Stortford. 20th January


Cafe Scientifique, Bishop's Stortford

Next event:

Monday January 20th 2014 **Doors open at 6:45 for a 7:15 start**

It's In Your Genes - What do you want to know?

Dr Anna Middleton




A saliva sample contains all 20,000 or so genes that every person has. It is now technically possible as well as relatively cheap and easy to look through all 20,000 and offer information about your past, present and future health. Genes contain information about when you would have been predisposed to developing your first tooth through to whether you are at risk of developing Alzheimers when you are older. What, if anything, would you want to know? This 'sequencing' technology is going to be in the NHS on a large scale from next year and in preparation for this Anna Middleton is running a social sciences research project that has gathered the attitudes from 7,000 people across 91 different countries on how they feel the testing should be implemented.

In this talk, Anna will give an overview of the science as well as how people are interested in using it. Members of the audience will also have the opportunity to see how they compare by using voting pads to register their attitudes.

Next Event:
Booking
Google Map of Venue
Alternative Map of Venue
List of Previous Events
Cafe-Sci.Org
Stortford on CafeSci.Org

If you get "Page Not Found", click one of the Navigation links above



4
days until
It's In Your Genes

Text too small?
If your Mouse has a Scroll Wheel, just hold the Control key and scroll the wheel to change the Font Size

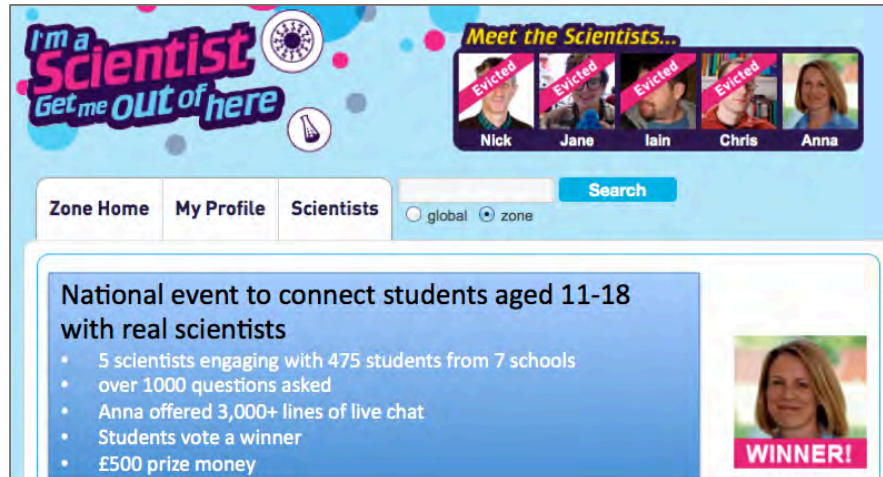
Middleton A (2014) Ethics and Genomics. Teaching on the Wellcome Trust Advanced Course for professionals: Fundamentals of Clinical Genetics, Genome Campus, Hinxton, Cambridge, 17th January

Middleton A (2013) Genomics, ethics: what's all the fuss about incidental findings? Sheffield Institute of Biotechnological Law and Ethics at Sheffield University, 27th November

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2014) winner on I'm a scientist get me out of here!, a Wellcome Trust supported national event to connect students aged 11-18 with real scientists, March 2014



Middleton A (2013) Genomics, ethics and what people want to know. Teaching on the Wellcome Trust Advanced Course for professionals: Molecular Pathology and Diagnosis of Cancer, Genome Campus, Hinxton, Cambridge, 15th November

Middleton A (2013) What's all the fuss about incidental findings? Genethics Club Plenary presentation. Addenbrooke's Hospital, Cambridge, 8th November

GENETHICS

www.genethicsclub.org

Genethics club is for health professionals- and other interested parties- to discuss and explore difficult ethical and or legal issues encountered in genetic medicine.

Next meeting: Cambridge
Friday 8th November, 2013
10.30am- 4.30pm

Venue: Meeting Rooms 6A & 6B, Level 6 The Addenbrookes Treatment Centre,
 Cambridge University Hospitals, Hills Road, Cambridge, CB2 0QQ www.cuh.org.uk
tel no for queries on the day: Dept of Clinical Genetics: 01223 216446

Plenary: "What's all the fuss about Incidental findings?"

Anna Middleton; GC and researcher,
 Wellcome Trust Sanger Institute, Cambridge

Please bring any clinical cases or issues for discussion.
All Welcome

A charge of £15 per head will be made for the day.
This will include lunch and refreshments.
If you would like to attend please contact:
 Mike Parker or Anneke Lucassen
michael.parker@ethox.ox.ac.uk
a.m.lucassen@soton.ac.uk

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2013) Genomics, ethics and what people want to know. Teaching on the University of Cambridge and Life Technologies training event: 'Genomics in Medicine' September 27th, approved by the Federation of the Royal Colleges of Physicians

Middleton A (2013) Tweeting, blogging and just plain winging it: utilising social media to recruit into social sciences research. HeLEX Centre for Health, Law and Emerging Technologies, University of Oxford, 9th September

Middleton A (2013) Genomics, ethics and what people want to know. British Humanist Society, Cambridge, 21st August

Middleton A, Patch C, Wiggins J, Barnes K, Crawford C, Benjamin C, Bruce A on behalf of the Association of Genetic Nurses and Counsellors in the UK (2013) Position Statement on Opportunistic Genomic Screening. AGNC website and publication forthcoming

Middleton A (2013) International attitudes towards sharing 'incidental findings' from whole genome research studies: empirical data from health professionals, genomic researchers and the public. Public Health Genomic Foundation on Realising Genomics in Clinical Practice. 9-10 July, Madingley Hall, Cambridge

Middleton A (2013) Ethics and Social Science. DDD Collaborators meeting. Hinxton Hall, Cambridge, 24th May

Middleton A (2013) Critique of recent ACMG recommendations on incidental findings. Workshop on human disease related variants. 22nd April. Sanger/EBI, Cambridge

Middleton A (2011) Ethical implications of genomics. Foundation Brocher, 11th August, Geneva, Switzerland

Middleton A (2011) Ethics, genomics and Who Wants to be a Millionaire. Collaborative Group for Genetics in Healthcare GenRes NIHR Genetics Specialty Group Conference 1st July, Newcastle

Middleton A (2011) Ethical aspects of whole genome analysis. 7th International DECIPHER symposium. 23-25 May, Cambridge

Invited Presentations, Seminars, Book Chapters and Teaching that Includes Work on the GenomEthics Study

(section 4 of 8)

Middleton A (2011) Genethics: what to do with incidental findings from whole genome studies? Invited seminar for the Human Genetics Group at the Wellcome Trust Sanger Institute, Cambridge, 19th April

Middleton A (2011) Genethics: whole genome studies, incidental findings and the DDD project. Invited seminar for the East Anglia Regional Genetics Service, Addenbrooke's Hospital, Cambridge, 14th April

Middleton A (2011) Invited panel member of Personal Genomics Debate at Cambridge Next Generation Sequencing Day, Centre for Mathematical Sciences at University of Cambridge 28th March

Middleton A (2011) The Ethics of Genetics: an exploratory study on the views of patients and health professionals involved in the DDD project. Invited seminar at the Wellcome Trust Sanger Institute, Cambridge, 18th Feb

Video, Museum Exhibits and Teaching Materials Based on Outcomes of the GenomEthics Study

(section 5 of 8)

Invited presentation given to the UK Biobank Ethics and Governance Council, 2014

(video screen captures, full video viewable at:

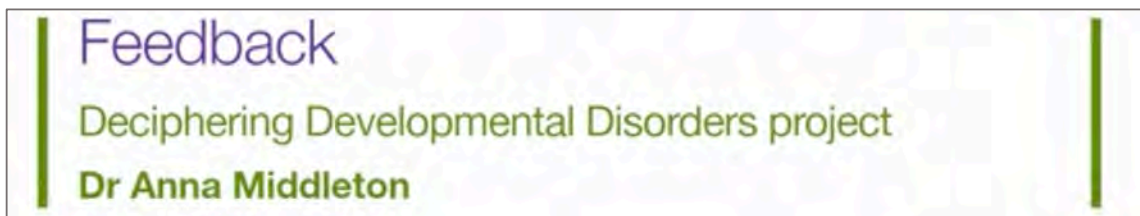
https://www.youtube.com/watch?time_continue=47&v=jGz02hCrrPo)



Past, present, future:
the ethics and governance
of big biobanks conference

4 November 2014
Wellcome Trust, 215 Euston Road, London

**UK Biobank
Ethics and
Governance
Council**



Feedback

Deciphering Developmental Disorders project

Dr Anna Middleton

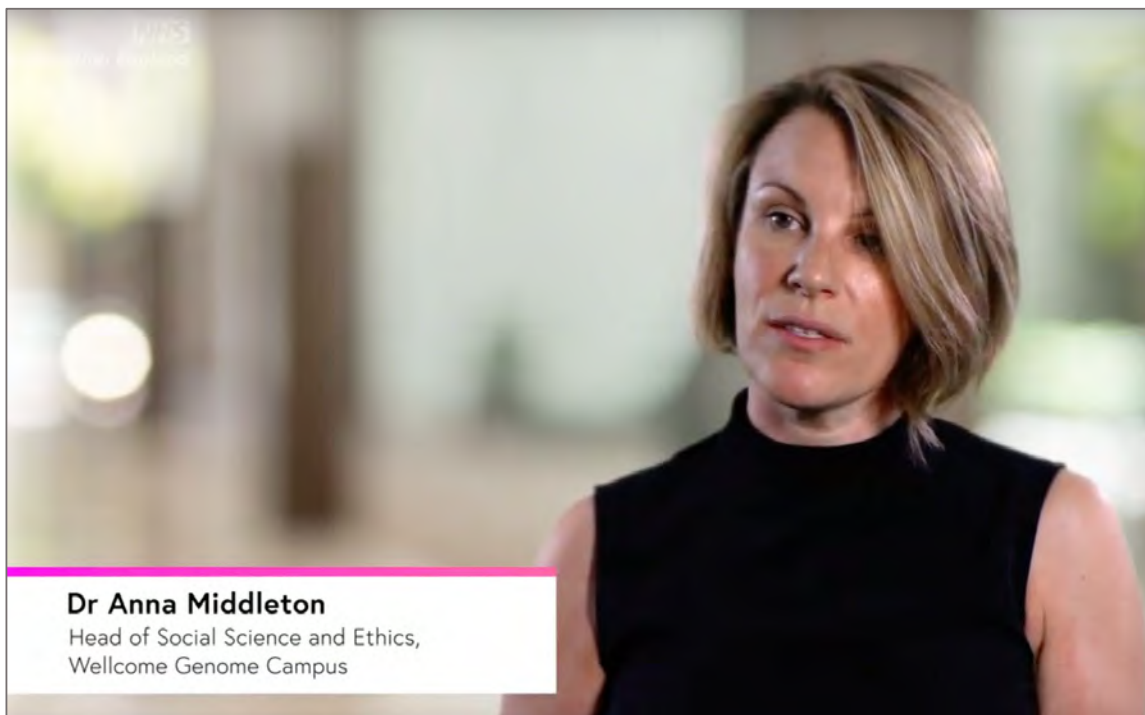


Video, Museum Exhibits and Teaching Materials Based on Outcomes of the GenomEthics Study

(section 5 of 8)

Invited Interview with Genomics Education Programme, Health Education England, 2016. Used for training health professionals

(video screen captures, full video viewable at: <https://www.youtube.com/watch?v=sKIMp4XAh7U>)



Video, Museum Exhibits and Teaching Materials Based on Outcomes of the GenomEthics Study

(section 5 of 8)

Interviews on ethics and genomics, 2014 for teaching on:

- NHS National Genetics and Genomics for Healthcare website
- Health Education England and NOWGEN e-learning course on bioinformatics 2014
- 7 x videos produced

(video screen captures, full videos viewable on playlist at: <https://vimeo.com/92617962>)

Reducing potential harm by managing expectations

People have different views about the amount and type of genomic information they want

Ethical principles relating to medical information also apply to genomic information

Confidentiality of large scale personal health data concerns many people

Deciding which genomic information to interpret involves multidisciplinary teams

Video, Museum Exhibits and Teaching Materials Based on Outcomes of the GenomEthics Study

(section 5 of 8)

Genomics Education Programme & Health Education England 2015
Course on Taking Consent for 100k Genomes Project

- Co-writer of core curriculum
- Co-Creator of 6 x videos produced as course teaching aids

(literature screen captures, full PDF document viewable at:

https://www.genomicseducation.hee.nhs.uk/images/pdf/HEEConsentCoursePDF_Dec15-final.pdf)

100,000 GENOMES PROJECT
Preparing for the consent conversation

Acknowledgements

This Health Education England course has been developed in collaboration with:

Writing and subject matter expertise: Dr Anna Middleton, Wellcome Trust Sanger Institute; Dr Christine Patch, Guy's and St Thomas'; Siobhan Chan, Progress Educational Trust; Dr Denise Williams, Dr Kai Ren Ong and colleagues at Birmingham Women's NHS Foundation Trust's Clinical Genetics department	Advisory group members: Dr Laura Boyes, Birmingham Women's NHSFT; Mary Elford, Health Education England; Dr Tom Fowler, Genomics England; Alison Hall, PHG Foundation; Stephanie Jones, John Radcliffe Hospital, Oxford; Jane Niederer, Churchill Hospital, Oxford; Dr Elizabeth Ormondroyd, University of Oxford; Prof Michael Parker, Genomics England; Vivienne Parry, Genomics England; Jessica Wright, University of Oxford	Genomics England: Dr Jeanna Mahon-Pearson, Laura Riley, Dr Clare Turnbull, Dr Ellen Thomas and colleagues	HEE Genomics Education Programme team: Dr Michelle Bishop, Sandie Gay, Aine Kelly, Michelle Madeley, Tim Packwood, Dr Anneke Seller, Stuart Sutherland, and colleagues
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Dr Anna Middleton
Ethics Researcher, Genetic Counsellor
Wellcome Trust Sanger Institute

Video, Museum Exhibits and Teaching Materials Based on Outcomes of the GenomEthics Study

(section 5 of 8)

The videos from the Genomethics survey were used by Genomics England on their website to help website visitors engage with ethical issues



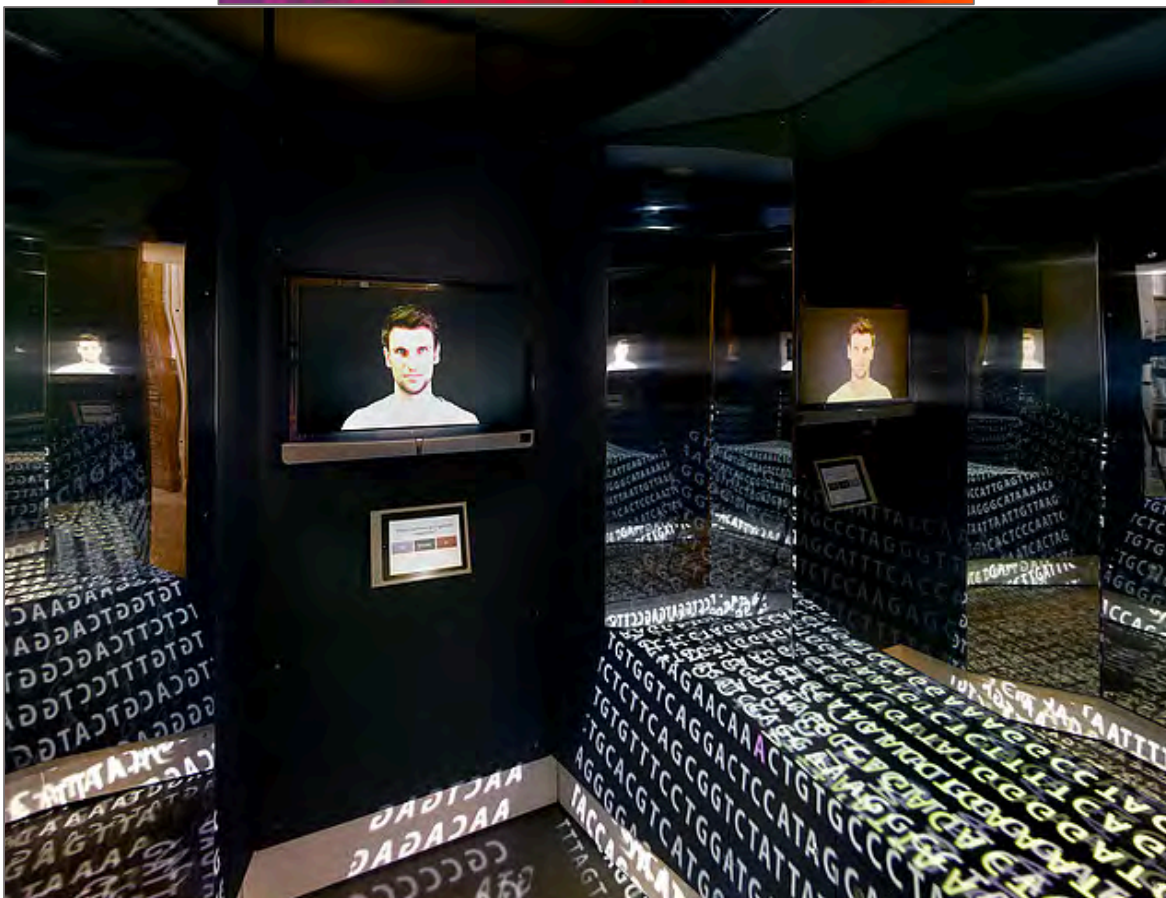
The Science Museum exhibit "How is Gene Editing Transforming Medicine?" Installed October 2016 using input from Anna Middleton's research.



Video, Museum Exhibits and Teaching Materials Based on Outcomes of the GenomEthics Study

(section 5 of 8)

Helped design the 'ethics' component of the Sanger stand at the Royal Society Festival of Science, July 2013, utilising a multi-media approach consisting of film with actors asking a series of ethics questions. This was delivered on a large TV screen and visitors could press buttons to answer the questions. Several projectors beamed down genetic 'traits' as words onto visitor's clothes. The exhibition was designed to run concurrently with a similar exhibition at the Smithsonian Museum in Washington DC which utilised the same video.



GENOMETHICS

DDD/Ethics Project

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News Coverage and Media Referring to the GenomEthics Study

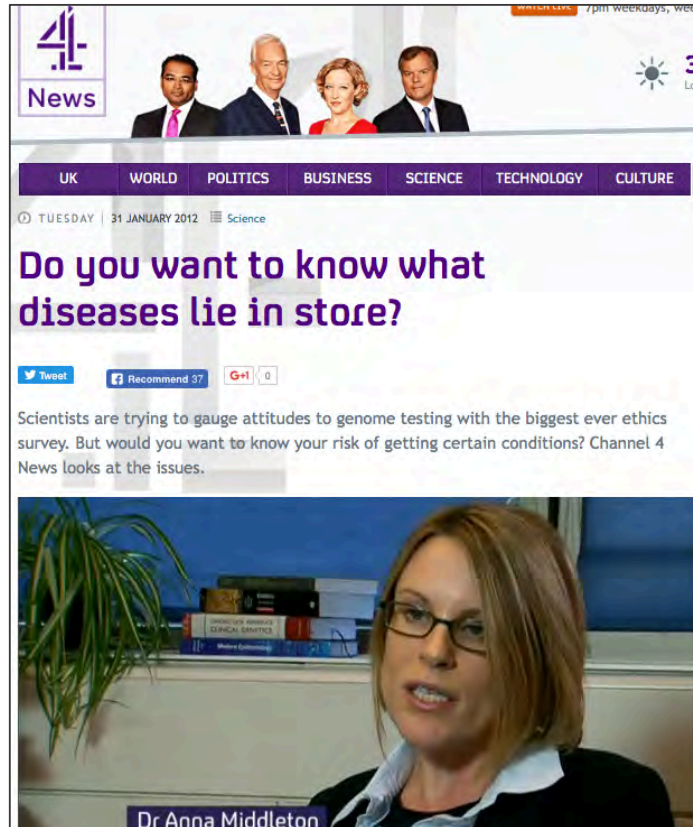
(section 6 of 8)

Channel 4 News, 31 January 2012

Mix media - Online article with video interview

(reader reaction captured 17 October 2016)

Do you want to know what diseases lie in store? - Presented by Krishnan Guru-Murthy



What do you think?

Get in touch on [Facebook](#) or via [Twitter @channel4news](#) and your thoughts could be featured below.

Valerie Callen: "I would not want to know. Why would I want to spend my whole life worrying about dying, which is inevitable anyway! I say, live and happy living!"

James Snowdon: "There's something a little bit 'playing god' about any kind of genetic profiling. How long until parents can decide to abort children because of a high diabetes risk? Frankly I'm all for this though - let's go the whole way and bring on designer babies and mechanical organs. Yay modern medicine."

John Hickman: "I'd love to know, so I can do all I can to avoid nasty conditions but I think there also needs to be very capable information available on the implications of findings when discovered, almost a counselling service."

Colin Robinson: "In my opinion this will no doubt come down to monetary terms in the long run...Insurance companies and care providers will use this information...I have no problem with people knowing what the risks for themselves are. They can use it to make informed decisions on lifestyle and parenting and so on. The real problem for me is whether we will be disadvantaged/prejudiced against because of the same information."

GENOMETHICS

DDD/Ethics Project

News Coverage and Media Referring to the GenomEthics Study

(section 6 of 8)

BBC Look East News, 1 February 2012

Mix media - Online article with video interview

Sanger Institute's big gene survey to test public's view - Presented by Mike Cartwright



BBC Sign in News Sport Weather iPlayer TV Radio

NEWS ENGLAND

Home World UK **England** N. Ireland Scotland Wales Business Politics Health Education Sci/Environ

Sanger Institute's big gene survey to test public's view



Dr ANNA MIDDLETON
BBC LOOK EAST

1 February 2012 Last updated at 12:40

A genetics survey has been set up online to find out how many people want to know what medical conditions they may face in the future.

The survey is being conducted by the Sanger Institute in Cambridge, which played a big part in mapping the human genetic code.

By examining DNA, doctors can tell if people are likely to develop medical conditions like diabetes, Alzheimer's and cancer. The institute is trying to establish whether or not people would want to know.

News Coverage and Media Referring to the GenomEthics Study (section 6 of 8)

Channel 4 News, 14 June 2013

DNA link to Prince William's Indian Ancestry by Asha Tanna

Breach of privacy?

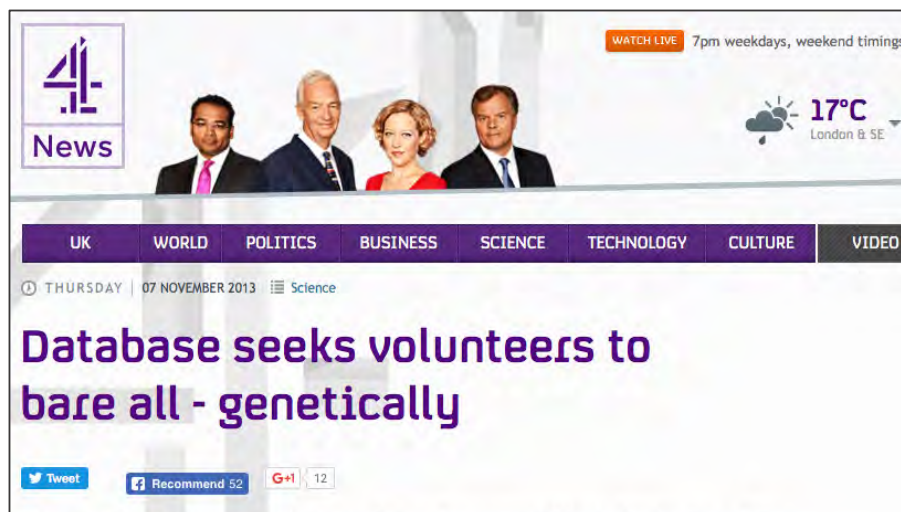
But the revelation which made the front page of the British newspaper the Times has been criticised by some in the field of genetics, who say not only is it a breach of privacy towards the prince, but also distasteful that the newspaper ran an advert of the company doing the testing at the same time.

Writing for the website The Conversation, Dr Anna Middleton, ethics researcher and genetic counsellor at the Wellcome Trust Sanger Institute, said: "Genetics unites all of us. But revealing personal information about somebody, without their consent, irrespective of their position or status, is potentially harmful.

"While the discussion is about ancestry today, it could be more serious if predispositions to life-threatening conditions are revealed? If it were me, I'd rather know this first and have a chance to talk to my family before anybody else knows about it."

Channel 4 News, 7 November 2013

Database seeks volunteers to bare all - genetically by Emma Maxwell



The image is a screenshot of the Channel 4 News website. At the top left is the Channel 4 logo and the word "News". To the right, there is a "WATCH LIVE" button and the text "7pm weekdays, weekend timings". Below this is a weather widget showing "17°C" and "London & SE". A navigation bar contains categories: UK, WORLD, POLITICS, BUSINESS, SCIENCE, TECHNOLOGY, CULTURE, and VIDEO. Below the navigation bar, the date and time are shown as "THURSDAY | 07 NOVEMBER 2013 | Science". The main headline reads "Database seeks volunteers to bare all - genetically". At the bottom of the headline area, there are social media sharing buttons for "Tweet", "Recommend 52", and "G+ 12".

News Coverage and Media Referring to the GenomEthics Study (section 6 of 8)

BBC Radio 4, 2 February 2012

Radio interview

Discussion of a survey of ethical attitudes to sharing genomic information from the show Material World featuring Quentin Cooper



BBC Radio Cambridgeshire, 17 April 2013

Radio interview

Introduction on research on Incidental Findings on Pail Stainton Bigger Breakfast Show



News Coverage and Media Referring to the GenomEthics Study

(section 6 of 8)

The Naked Scientists, (2015). [Radio programme] 97.2: Cambridge University Radio

Genetics and Ethics interview from the show

Interviewed by Dr. Kat Arney for the show A hundred thousand genomes



The screenshot shows the 'The Naked Scientists' website interface. At the top left is a logo featuring a stylized figure with four arms in a white lab coat against a yellow background. To the right of the logo is the text 'THE NAKED SCIENTISTS' and the University of Cambridge crest and name. The main heading is 'Science Interviews'. Below this is a navigation menu with categories: Home, Podcasts, Science News, Experiments, Articles, Materials Science, Science Forum, Interviews (selected), Biology, Chemistry, Medicine, Nature, Physics, Space & Earth, and Other. The main content area displays an interview titled 'Anna Middleton - Genetics and ethics' dated 'TUE, 10TH MAR 2015'. The guest is identified as 'Anna Middleton, Wellcome Trust Sanger Institute'. There are links for 'Listen Now' and 'Download as mp3 from the show A hundred thousand genomes'. A short excerpt of the interview text is visible at the bottom.

The Naked Scientists, (2015). [Radio programme] 97.2: Cambridge University Radio

Do you want to know what's in your DNA?

Interviewed by Chris Smith for the show Safety at 40,000 Feet



The screenshot shows the 'The Naked Scientists' website interface. At the top left is a logo featuring a stylized figure with four arms in a white lab coat against a yellow background. To the right of the logo is the text 'THE NAKED SCIENTISTS' and the University of Cambridge crest and name. The main heading is 'Science Interviews'. Below this is a navigation menu with categories: Home, Podcasts, Science News, Experiments, Articles, Materials Science, Science Forum, Interviews (selected), Biology, Chemistry, Medicine, Nature, Physics, Space & Earth, and Other. The main content area displays an interview titled 'Do you want to know what's in your DNA?' dated 'MON, 11TH MAY 2015'. The guest is identified as 'Anna Middleton, Sanger Institute'. There are links for 'Listen Now' and 'Download as mp3 from the show Safety at 40,000 Feet'.

GENOMETHICS

DDD/Ethics Project

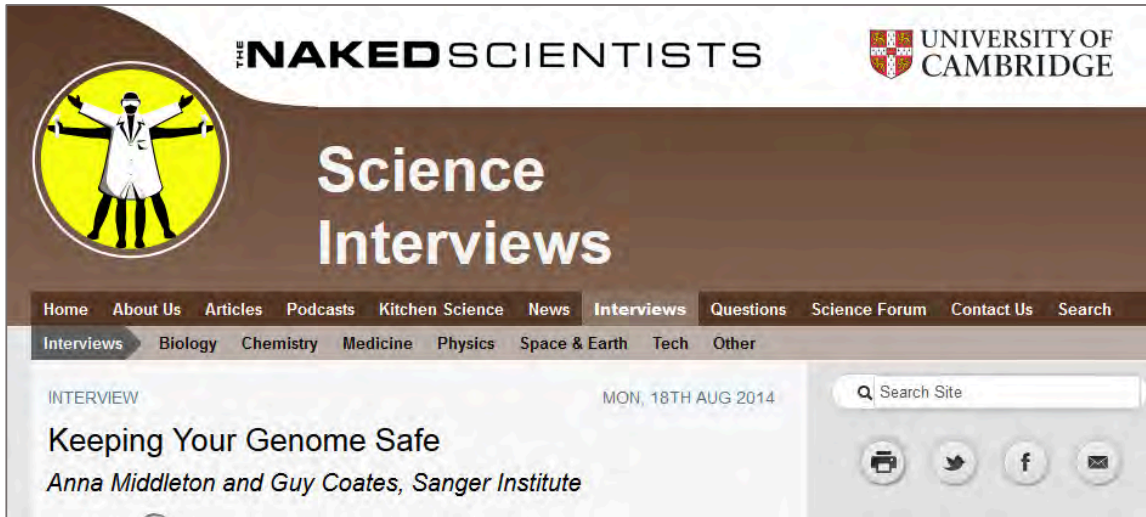
News Coverage and Media Referring to the GenomEthics Study

(section 6 of 8)

The Naked Scientists, (2014). [Radio programme] 97.2: Cambridge University Radio

Keeping your genome safe

Interviewed (along with Guy Coates) by Graihagh Jackson for the show *The Future of Medicine*



Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Ratchinsky, K. (2016). What Digital Innovation in Health Care Will Look Like. *Observer* [online]. Available at <http://observer.com/2016/05/what-digital-innovation-in-healthcare-will-look-like/> [Accessed 16 Nov. 2016]

OBSERVER POLITICS | ARTS & ENTERTAINMENT | STYLE & DESIGN | BUSINESS

What Digital Innovation in Health Care Will Look Like

By [Karin Ratchinsky](#) • 05/02/16 10:21am

[f](#) [t](#) [in](#) [G+](#) [e](#)



Digitization of healthcare records has literally paved the way for a far more informed, innovative, and personalized care paradigm. (Photo: Wesley Wilson/Pexels)

In the growing area of personalized medicine, even healthy people have a strong interest in their susceptibility to disease now that genomic research has scaled down economically to the individual level. A study done by the *European Journal of Human Genetics* reports 98 percent of stakeholders in a nearly 7,000-person research cohort wanted to know about preventable life-threatening conditions revealed through genetic screening.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Lewis, R (2013). Why 23andM is Not for Met - Yet. *PLOS Blogs: DNA Science Blog - Genetics in Context* [online]. Available at <http://blogs.plos.org/dnascience/2013/11/27/why-23andme-is-not-for-me-yet/> [Accessed 19 Oct. 2016]



A LESSON FROM THE "INCIDENTAL FINDINGS" SITUATION?

At the ASHG annual meeting in Boston last month, a wonderful talk by Anna Middleton, PhD, a research associate at the Wellcome Trust Sanger Institute in Cambridge, UK, confirmed what I'd noticed about my colleagues. Her work as an ethics researcher and genetic counselor has shown that people's feelings about how much they want to know about their DNA tracks not so much with age, religion, income level or geography, but with profession.

The talk was part of a session on incidental findings: having a test to diagnose one condition, and finding unexpected evidence of another. (See "Incidental findings from genome sequencing: nuances and caveats" from [Scientific American blogs](#).) Direct-to-consumer testing, of course, isn't the same as a research study in a clinical setting; multiple findings are intended, not incidental. But the issue of how to handle complex genetic information is similar.



(Dept. of Energy)

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

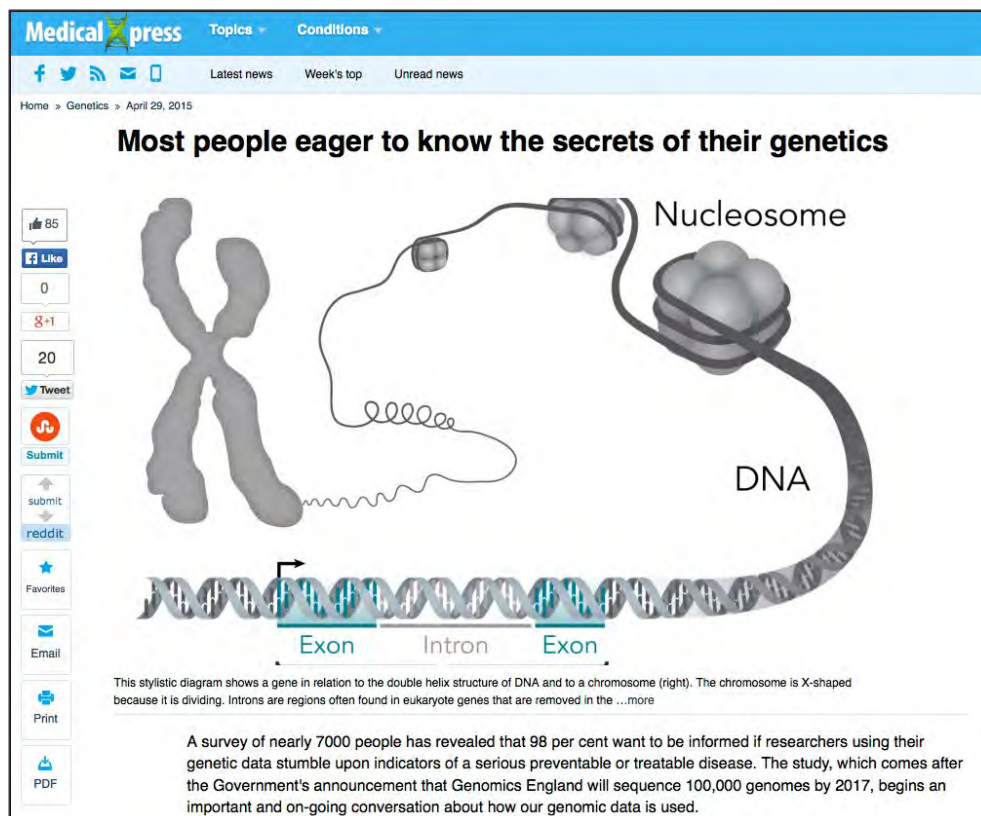
Wellcome Trust Sanger Institute (2015). Most people eager to know the secrets of their genetics. Picked up by the following sources:

15 Minute News [online]. [Accessed in 2015]



The screenshot shows the top of a news website. At the top left is the '15 MINUTE NEWS' logo. To the right are navigation links for 'Headlines', 'Business', 'Sports', 'Entertainment', 'Technology', and 'Style'. Below these are two featured article teasers: 'Climber retrieves friend's...' from CNN (dated 5 May 2015 03:52) and 'Jennifer Lopez's Met Gala gown...' from Daily Mail (dated 5 May 2015 06:21). The main article is titled 'Most people eager to know the secrets of their genetics' from Science Daily, dated 29 Apr 2015 14:51. The article text begins: '98 per cent of people want to be told about indicators of a serious preventable or treatable disease found by researchers using their genomic data, according to a survey of nearly 7,000 people. Currently, a lot of the information in our personal genomes is of uncertain clinical significance. As genetic research advances and more can be learned from sequence data, this survey will help researchers and health policy makers to plan accordingly.'

Medical Xpress [online]. Available at <http://medicalxpress.com/news/2015-04-people-eager-secrets-genetics.html#nRlv> [Accessed 14 Nov. 2016]



The screenshot shows a news article on the Medical Xpress website. The article title is 'Most people eager to know the secrets of their genetics'. On the left side, there is a vertical sidebar with social media sharing options: Like (85), 0, +1, 20, Tweet, and other buttons. The main content area features a diagram illustrating the relationship between a chromosome, DNA, and a gene. The chromosome is shown as an X-shaped structure on the left. A line representing DNA extends from the chromosome to the right, where it is shown as a double helix. A section of the DNA double helix is highlighted in blue and labeled 'DNA'. Below this section, the structure is further detailed, showing 'Exon' and 'Intron' regions. A 'Nucleosome' is also labeled, showing the DNA wrapped around a protein core. Below the diagram, there is a caption: 'This stylistic diagram shows a gene in relation to the double helix structure of DNA and to a chromosome (right). The chromosome is X-shaped because it is dividing. Introns are regions often found in eukaryote genes that are removed in the ...more'. At the bottom of the article, there is a paragraph of text: 'A survey of nearly 7000 people has revealed that 98 per cent want to be informed if researchers using their genetic data stumble upon indicators of a serious preventable or treatable disease. The study, which comes after the Government's announcement that Genomics England will sequence 100,000 genomes by 2017, begins an important and on-going conversation about how our genomic data is used.'

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Wellcome Trust Sanger Institute (2015). Most people eager to know the secrets of their genetics.
Picked up by the following sources (*continued*):

Science Codex [online]. Available at
http://www.sciencecodex.com/most_people_eager_to_know_the_secrets_of_their_genetics-156171 [Accessed 30 Apr. 2015]



The screenshot shows the top section of the Science Codex website. On the left, the 'SCIENCE CODEX' logo is displayed. To the right, there is a promotional banner for Avon: 'Become an Independent Avon Representative in your local area' with an image of Avon hairbrushes and a bottle of 'Gel Softy' hair gel. Below the banner is a navigation menu with tabs for 'Home', 'Earth', 'Heavens', 'Body', and 'Brain'. The main article title is 'Most people eager to know the secrets of their genetics', posted on April 29, 2015, at 9:30pm. Social sharing buttons for '+ Share / Save', Facebook, Twitter, Google+, and LinkedIn are visible. The article text begins: 'A survey of nearly 7,000 people has revealed that 98 percent want to be informed if researchers using their genetic data stumble upon indicators of a serious preventable or treatable disease. The study, which comes after the Government's announcement that Genomics England will sequence 100,000 genomes by 2017, begins an important and on-going conversation about how our genomic data is used.'

ScienceDaily [online]. Available at
<https://www.sciencedaily.com/releases/2015/04/150429085136.htm>
[Accessed on 14 Nov. 2016]



The screenshot shows the ScienceDaily website interface. At the top left is the 'ScienceDaily' logo with the tagline 'Your source for the latest research news'. On the right, there are 'Mobile' links for iPhone and Android, and a 'Breaking News: Lumino' alert. A large advertisement for Sky+HD boxes is featured, stating 'Get a free Sky+HD box when you join Sky TV online'. Below the ad is a navigation menu with categories: 'Health', 'Physical/Tech', 'Environment', 'Society/Education', and 'Quirky'. The main article title is 'Most people eager to know the secrets of their genetics', with the source 'from research organizations'. The article details include: 'Date: April 29, 2015', 'Source: Wellcome Trust Sanger Institute', and a 'Summary' stating: '98 per cent of people want to be told about indicators of a serious preventable or treatable disease found by researchers using their genomic data, according to a survey of nearly 7,000 people. Currently, a lot of the information in our personal genomes is of uncertain clinical significance. As genetic research advances and more can be learned from sequence data, this survey will help researchers and health policy makers to plan accordingly.'

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Wellcome Trust Sanger Institute (2015). Most people eager to know the secrets of their genetics. Picked up by the following sources (*continued*):

Bioethics.com [online]. Available at

<http://www.bioethics.com/archives/27197> [Accessed 14 Nov. 216]

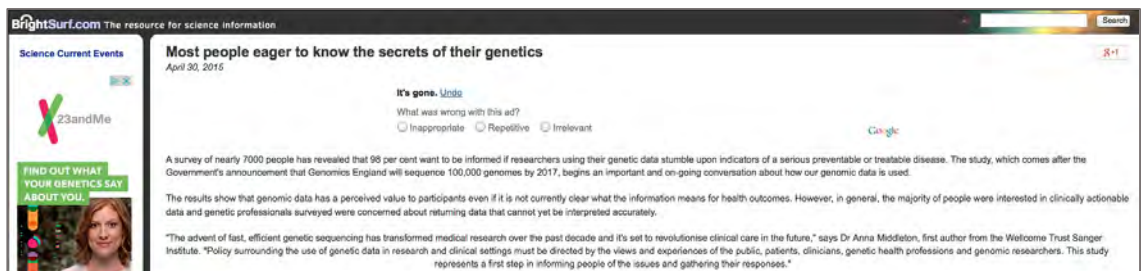


(e)Science News [online]. Available at

<http://esciencenews.com/sources/science.daily/2015/04/29/most.people.eager.know.secrets.their.genetics> [Accessed 14 Nov. 216]



BrightSurf.com [online]. [Accessed in 2015]



Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Wellcome Trust Sanger Institute (2015). Most people eager to know the secrets of their genetics. Picked up by the following sources (*continued*):

HealthBZ [online]. Available at <https://healthbz.wordpress.com/2015/04/29/most-people-eager-to-know-the-secrets-of-their-genetics/> [Accessed on 14 Nov. 2016]



The screenshot shows a HealthBZ article. On the left is a photo of a woman in a green tank top sitting cross-legged, holding a bunch of vegetables and a purple dumbbell. Below the photo is the HealthBZ logo and the text 'Get the latest health & wellness advice'. To the right of the photo, it says 'Posted on April 29, 2015 | Today's Healthcare News -- ScienceDaily'. The main title is 'Most people eager to know the secrets of their genetics'. Below the title is a sub-headline: '98 per cent of people want to be told about indicators of a serious preventable or treatable disease found by researchers using their genomic data, according to a survey of nearly 7,000'.

Cambridge Network [online]. Available at <http://www.cambridgenetwork.co.uk/news/most-people-eager-to-know-the-secrets-of-their-genetics/> [Accessed on 14 Nov. 2016]



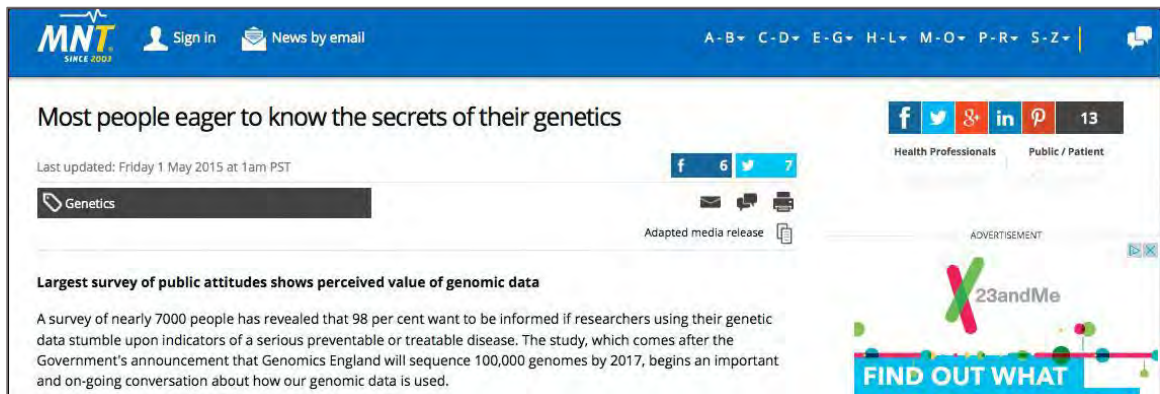
The screenshot shows a Cambridge Network article. At the top left is the Cambridge Network logo. To the right are navigation links: Home, News, Jobs, Events, Learning, Directory. Below the logo is a breadcrumb trail: Cambridge Network > Home > News > Most people eager to know the secrets of their genetics. The main title is 'Most people eager to know the secrets of their genetics'. Below the title is the date '30/04/2015'. To the left of the main content are four sidebar links: 'Newsletter' (with a laptop icon), 'News Archive' (with a stack of papers icon), 'In the press' (with a newspaper icon), and 'Founder views' (with a silhouette icon). Below these links is a 'Newsletter signup' form with an 'Email address' input field and a 'Go' button. The main content area features a large image of a blue DNA double helix. Below the image is the text: 'A survey of nearly 7000 people has revealed that 98 per cent want to be informed if researchers using their genetic data stumble upon indicators of a serious preventable or treatable disease. The study, which comes after the Government's announcement that Genomics England will sequence 100,000 genomes by 2017, begins an important and on-going conversation about how our genomic data is used.'

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

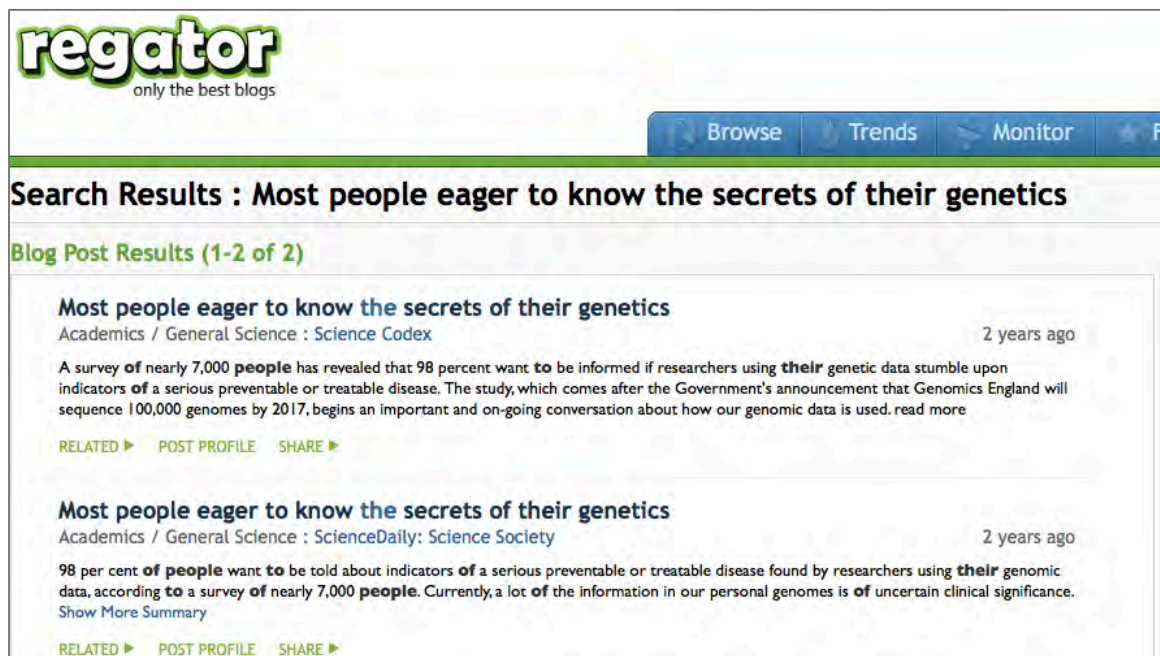
Wellcome Trust Sanger Institute (2015). Most people eager to know the secrets of their genetics. Picked up by the following sources (*continued*):

Medical News Today MNT [online]. Available at <http://www.medicalnewstoday.com/releases/293276.php> [Accessed on 14 Nov. 2016]



The screenshot shows the Medical News Today (MNT) website interface. At the top, there is a navigation bar with the MNT logo, a 'Sign in' button, and a 'News by email' button. A secondary navigation bar contains a list of letters: A-B, C-D, E-G, H-L, M-O, P-R, S-Z. The main content area features the article title 'Most people eager to know the secrets of their genetics' and a sub-headline 'Largest survey of public attitudes shows perceived value of genomic data'. The article text begins with 'A survey of nearly 7000 people has revealed that 98 per cent want to be informed if researchers using their genetic data stumble upon indicators of a serious preventable or treatable disease...'. To the right, there is a social media sharing section with icons for Facebook, Twitter, Google+, LinkedIn, and Pinterest, along with a '13' count. Below this is an advertisement for 23andMe with the text 'FIND OUT WHAT'.

Regator Only the Best Blogs [online]. Available at <http://regator.com/search/Most+people+eager+to+know+the+secret+s+of+their+genetics/> [Accessed on 14 Nov. 2016]



The screenshot shows the Regator search results page. The Regator logo is at the top left, with the tagline 'only the best blogs'. A navigation bar includes 'Browse', 'Trends', and 'Monitor'. The search results are for the query 'Most people eager to know the secrets of their genetics'. Two results are shown under 'Blog Post Results (1-2 of 2)'. The first result is from 'Academics / General Science : Science Codex' and is dated '2 years ago'. The second result is from 'Academics / General Science : ScienceDaily: Science Society' and is also dated '2 years ago'. Both results include a brief summary of the article and options for 'RELATED', 'POST PROFILE', and 'SHARE'.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

News Staff. 98 Percent Want to Know if Genetic Data Contains a Serious Preventable or Treatable Disease. *Science 2.0* [online]. Available at http://www.science20.com/news_articles/98_percent_want_to_know_if_genetic_data_contains_a_serious_preventable_or_treatable_disease-155224 [Accessed in 15 Nov. 2016]



News Staff. 98 Percent Want to Know if Genetic Data Contains a Serious Preventable or Treatable Disease. Picked up by the following source:

(e) *News Science* [online]. Available at <http://esciencenews.com/sources/scientific.blogging/2015/04/29/98.percent.want.to.know.if.genetic.data.contains.a.serious.preventable.or.treatable.disea> [Accessed on 15 Nov. 2016]



Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

MacDonald, G. (2015). My DNA, my data: British people would want to know if they have genes linked to diseases. *BioPharma-Reporter* [online]. Available at <http://www.biopharma-reporter.com/Bio-Developments/My-DNA-my-data-British-people-would-want-to-know-if-they-have-genes-linked-to-diseases> [Accessed 14 Nov. 2016]

The screenshot shows a webpage from BioPharma-Reporter.com. The header includes the site logo, 'FREE NEWS', and a navigation menu with links for HEADLINES, HOT TOPICS, RESOURCE LIBRARY, EVENTS, ON THE ROAD, and RELATED SITES. A red banner encourages registration for a 'FREE BioPharma-Reporter Fermentation Science &...' event. Below the navigation is a 'HEADLINES > BIO DEVELOPMENTS' section with text size, print, and forward icons, and social media sharing buttons for Facebook, Twitter, Google+, LinkedIn, and a plus sign, along with a '34' comment count. A subscription box prompts users to 'Subscribe to our FREE newsletter' with an email input field and a 'SUBSCRIBE' button. The main article title is 'My DNA, my data: British people would want to know if they have genes linked to diseases' by Gareth MacDonald, dated 29-Apr-2015. A large image of a colorful DNA microarray is featured, with a caption: 'My DNA, My data: Brits would want disease-related genomic info say researchers'. Below the image are related tags: 'Drug developmnet, Rare diseases, Genomic, Genetic, Information, UK, Wellcome Trust, DoH'. A sub-headline reads: 'Most Brits would want to know if they carry genes linked to life-threatening diseases according to UK researchers who say this could have implications for drug development.' The text states the finding is from a UK Department of Health (DoH) and Wellcome Trust funded survey of nearly 7,000 people. A logo for 'CMC biologics' is shown. The text continues: 'The researchers found that 98% of respondents said they would want access to their genomic data if it related to potential diseases, even if the risk was relatively low.' The lead researcher, Anna Middleton, is mentioned as having told Bio-Pharmareporter.com 'We'.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Middleton, A (2015). Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research. Picked up by the following source: *BioPortfolio* [online]. Available at <http://www.bioportfolio.com/news/article/2314314/Attitudes-of-nearly-7000-health-professionals-genomic-researchers-and-publics-toward-the.html> [Accessed 17 Nov. 2016]



The image shows a banner for BioPortfolio. On the left, the BioPortfolio logo is displayed in white on a blue background, with the tagline "Biotech, Healthcare and Medical Resources" below it. To the right, a green box contains the text "Get a .com from the world's largest domain provider." Below the logo, the word "Advertisement" is written in small text above a yellow rectangular placeholder. To the right of this placeholder, the main headline of the article is written in blue: "Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research".

Press Association (2015). DNA feedback 'no one size fits all' *Press Association* [online]. [Accessed on 30 Apr. 2016]



The image is a screenshot of a news article snippet. The headline is "DNA feedback 'no one size fits all'" in bold black text. Below the headline is a sub-headline: "A vast majority of people around the world would like to be told if their genetic data indicates serious preventable or treatable disease, research has shown." Below the sub-headline is a social media sharing bar with icons for Facebook, Twitter, Google+, Email, and a plus sign, followed by a "0" comment count. Below the sharing bar is a photograph of a microscope with a blue light filter. At the bottom left, the "PRESS ASSOCIATION" logo is visible, along with the text "Last updated: 30 April 2015, 00:20 BST". At the bottom right, there is a "Print this story" button.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

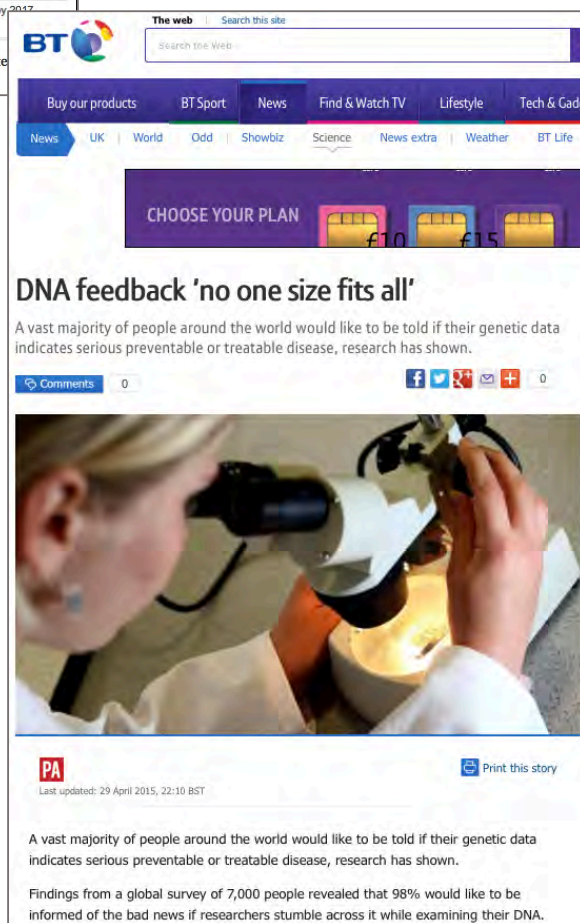
Press Association (2015). DNA feedback 'no one size fits all'. Picked up by the following sources:



The screenshot shows the Yahoo! News Digest UK & Ireland website. The main headline is "DNA feedback 'no one size fits all'" from the Press Association, dated Wednesday, April 29, 2015. Below the headline is a photograph of a person in a white lab coat looking through a microscope. The article text is partially visible, starting with "A vast majority of people around the world would like to be told if their genetic data indicate preventable or treatable disease, research has shown."

Yahoo! News Digest UK & Ireland [online]. [Accessed on 29 Apr. 2016]

BT.com News and Science [online]. Available at <http://home.bt.com/news/science-news/dna-feedback-no-one-size-fits-all-11363978484178> [Accessed on 14 Nov. 2016]



The screenshot shows the BT.com News and Science website. The main headline is "DNA feedback 'no one size fits all'" from the Press Association. Below the headline is a photograph of a person in a white lab coat looking through a microscope. The article text is partially visible, starting with "A vast majority of people around the world would like to be told if their genetic data indicates serious preventable or treatable disease, research has shown."

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Press Association (2015). DNA feedback 'no one size fits all'. Picked up by the following sources (continued):

Jersey Evening Post [online]. [Accessed on 30 Apr. 2015]



TimesOfMalta.com [online]. Available at <http://www.timesofmalta.com/articles/view/20150501/health-fitness/DNA-feedback-no-one-size-fits-all-.566337> [Accessed on 14 Nov. 2015]

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Friday, May 1, 2015, 00:01 by PA

DNA feedback 'no one size fits all'

A vast majority of people around the world would like to be told if their genetic data indicates serious preventable or treatable disease, research has shown.

Findings from a global survey of 7,000 people revealed that 98 per cent would like to be

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Smith, C. (2015). Data Shows People Want to Know What's in Their Genome. *Front Line Genomics* [online]. Available at <http://www.frontlinegenomics.com/news/937/data-shows-people-want-to-know-whats-in-their-genome/> [Accessed 14 Nov. 2016]

Front Line Genomics

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Data Shows People Want To Know What's In Their Genome

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30 April 2015

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Results of the world's largest survey of public attitudes towards genomic data have been published.

The [European Journal of Human Genetics](#) has published a study on attitudes towards the return of incidental results from sequencing research. The discussion around the reporting of incidental findings has gathered momentum recently with the advancement of high profile, large scale, sequencing projects.

The survey was part of the work being carried out by the Deciphering Developmental Disorders (DDD) project, based out of the Wellcome Trust's Sanger Institute. From approximately 7000 participants, 98% stated that they wanted to be informed if researchers came across incidental findings of clinically actionable conditions.

"The advent of fast, efficient genetic sequencing has transformed medical research over the past decade and it's set to revolutionise clinical care in the future," says Dr Anna Middleton, first author from the Wellcome Trust Sanger Institute. "Policy surrounding the use of genetic data in research and clinical settings must be directed by the views and experiences of the public, patients, clinicians, genetic health professions and genomic researchers. This study represents a first step in informing people of the issues and gathering their responses."

GENOMETHICS

DDD/Ethics Project

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)



Brice, P. (2015). Public want to know medical findings from genomic research. *phg* [online]. Available at <http://www.phgfoundation.org/news/16674/> [Accessed 14 Nov. 2016]

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Public want to know medical findings from genomic research

Dr Philippa Brice
Thursday, 30 April 2015



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- Consultation
- Genetic Privacy & Consent
- Issues
- Post-Genomic Projects

A major international survey of almost 7,000 people has revealed significant differences in the view of potential genomic research participants and genetic health professionals on feedback of incidental findings (IFs).

IFs are unexpected but medically significant results that occur during investigations for research or clinical purposes. They are by no means unique to genomic analysis, but with the relatively recent advent of whole genome sequencing, they are a significant issue both for large-scale genomic research and clinical implementation.

Published in the *European Journal of Human Genetics*, the survey is the work of researchers at the Wellcome Trust Sanger Institute, and linked to the **Deciphering Developmental Disorders (DDD) project**. Respondents fell into four different categories: genomic researchers, genetic health professionals, non-genetic health professionals, and the public. Significant differences were observed between the views of people from these different categories, although not between those of people from different countries. The views of the public and genetics health-professionals showed the greatest divergence.

Whilst 98% of the public wanted to know about the possibility of preventable, life-threatening disorders, genetic health professionals were five times more likely to think that incidental findings should not be reported. The researchers say that this disconnect should be explored and addressed by researchers and clinicians, and taken into account in future policy development.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Care, A. (2015). Would you want to know if you carried a rare life-threatening genetic condition? *Cambridge News* [online]. [Accessed on 2 May 2015]



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Would you want to know if you carried a rare life-threatening genetic condition?

By **Cambridge News** | Posted: May 02, 2015 By Adam Care



How much should we be told about what's in our DNA?

Comments (1)

If you carried a life-threatening condition would you really want to know about it?

With advances in genomic research unlocking more and more of the secrets of DNA, the prospect of a complete breakdown of an individual's genome is moving ever closer, raising major questions about whether or not patients should be told what [doctors](#) can discover hidden in their genes.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

GenNews (2015). Study Shows People Want Access to Their Own Genetic Data - No Matter What. *GEN: Genetic Engineering & Biotechnology News* [online]. Available at <http://www.genengnews.com/gen-news-highlights/study-shows-people-want-access-to-their-own-genetic-data-no-matter-what/81251356> [Accessed on 15 Nov. 2016]



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GEN News Highlights

June 8, 2015

Study Shows People Want Access to Their Own Genetic Data—No Matter What



Click Image To Enlarge +

Large numbers of people say they want to see their genomic information even when the data are not health-related or are simply raw. [iStock/Drs_Schwartz]

The largest study to date of attitudes towards the use of genomic information shows that the majority of people want access to results from genome sequencing, even if these are not directly related to the condition for which the analysis has been undertaken. This applies even when the data are not health-related or are simply raw, says Anna Middleton, Ph.D., a principal staff scientist at the Wellcome Trust Sanger Institute. She is presenting the results of the survey at the annual conference of the European Society of Human Genetics today in Glasgow.

The survey was aimed at probing the attitudes of the various groups involved in sequencing research, including patients, the public, health professionals, and genomic researchers, towards the types of genomic information they would be interested in receiving. Just under

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Pedersen, T. (2015). Most People Want to Know Secrets of Their Genetics. *PsychCentral* [online]. Available at <http://psychcentral.com/news/2015/05/02/most-people-want-to-know-secrets-of-their-genetics/84171.html> [Accessed 14 Nov. 2016]

The screenshot shows the PsychCentral website interface. At the top, there is a navigation bar with links for Home, Conditions, Quizzes, News & Experts, Research & Resources, Find Help, and Pro. Below the navigation bar is a banner for Silversea Senior Cruises. The main content area features the article title "Most People Want to Know Secrets of Their Genetics" by Traci Pedersen, with a "2 min read" indicator. The article text discusses a survey of 75 countries showing that 98% of people would like to be informed of their genetic risk. It also mentions the Wellcome Trust Sanger Institute and Alastair Kent OBE. A sidebar on the left lists various topics under the "News" category, including Abuse, Addiction, ADHD, and many others. An image of a hand holding a tablet displaying a DNA helix is positioned to the right of the article text.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Pedersen, T. (2015). Most People Want to Know Secrets of Their Genetics. Picked up by the following source:

European Journal of Medical Genetics [online]. [Accessed on 2 May 2015]



European Society of Human Genetics press release (2015). People want access to their own genomic data, even when uninterpretable. *European Society of Human Genetics* [online]. Available at <https://www.eshg.org/13.0.html> [Accessed on 15 Nov. 2016]



ESHG 2015 Conference Press Release: People want access to their own genomic data, even when uninterpretable 03 Jun. 2015

Glasgow, United Kingdom: The largest study to date of attitudes towards the use of genomic information shows that the majority of people want access to results from genome sequencing, even if these are not directly related to the condition for which the analysis has been undertaken. This applies even when the data are not health-related or are simply 'raw', a researcher will tell the annual conference of the European Society of Human Genetics today (Monday).

Dr Anna Middleton, a Principal Staff Scientist at the Wellcome Trust Sanger Institute, Cambridge, UK, will describe the results of a survey into the attitudes of the various groups involved in sequencing research – patients, public, health professionals, and genomic researchers – towards the types of genomic information they would be interested in receiving. Just under 7000 people from 75 countries took part in an on-line survey, advertised on social and traditional media, and by an email list-serve.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

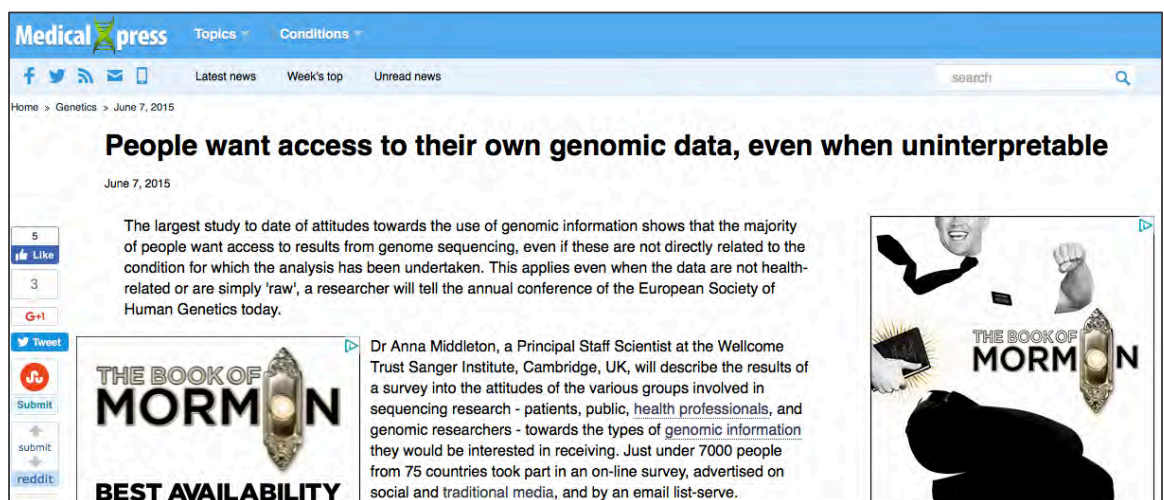
European Society of Human Genetics press release (2015). People want access to their own genomic data, even when uninterpretable. Picked up by the following sources:

EurekAlert! The Global Source for Science News [online]. Available at https://www.eurekalert.org/pub_releases/2015-06/esoh-pwa060415.php [Accessed on 15 Nov. 2016]



The screenshot shows the top of the EurekAlert! website. The header is red with the logo and the text 'The Global Source for Science News'. Below the header is a navigation bar with links for HOME, NEWS, MULTIMEDIA, MEETINGS, PORTALS, and ABOUT. The main content area features a public release dated 7-JUN-2015. The article title is 'People want access to their own genomic data, even when uninterpretable', attributed to the European Society of Human Genetics. There are social media sharing icons for Facebook, Twitter, LinkedIn, and YouTube, along with a 'SHARE' button. On the right, there are icons for 'PRINT' and 'E-MAIL'. The beginning of the article text is visible: 'Glasgow, United Kingdom: The largest study to date of attitudes towards the use of genomic information shows that the majority of people want access to results from genome'.

Medical Xpress [online]. [Accessed on 7 June 2015]



The screenshot shows the Medical Xpress website. The header is blue with the logo and navigation links for 'Topics' and 'Conditions'. Below the header is a navigation bar with links for 'Latest news', 'Week's top', and 'Unread news', along with a search bar. The main content area features a news article titled 'People want access to their own genomic data, even when uninterpretable', dated June 7, 2015. The article text states: 'The largest study to date of attitudes towards the use of genomic information shows that the majority of people want access to results from genome sequencing, even if these are not directly related to the condition for which the analysis has been undertaken. This applies even when the data are not health-related or are simply 'raw', a researcher will tell the annual conference of the European Society of Human Genetics today.' To the right of the text is an advertisement for 'THE BOOK OF MORMON' featuring a person holding a book. On the left side of the article, there are social media sharing icons for Facebook (5 likes), Twitter (3 tweets), LinkedIn, and Reddit.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

European Society of Human Genetics press release (2015). People want access to their own genomic data, even when uninterpretable. Picked up by the following sources (*continued*):

ScienceDaily [online]. Available at <https://www.sciencedaily.com/releases/2015/06/150607214204.htm> [Accessed on 15 Nov. 2016]

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Science News *from research organizations*

People want access to their own genomic data, even when uninterpretable

Date: June 7, 2015

Source: European Society of Human Genetics (ESHG)

Summary: The largest study to date of attitudes towards the use of genomic information shows that the majority of people want access to results from genome sequencing, even if these are not directly related to the condition for which the analysis has been undertaken. This applies even when the data are not health-related or are simply 'raw', a researcher states.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

European Society of Human Genetics press release (2015). People want access to their own genomic data, even when uninterpretable. Picked up by the following sources (*continued*):

Science Newsline Medicine [online]. Available at <http://www.sciencenewline.com/news/2015060813530013.html> [Accessed on 15 Nov. 2016]



SciFeeds [online]. Available at <https://scifeeds.com/news/people-want-access-to-their-own-genomic-data-even-when-uninterpretable/> [Accessed on 15 Nov. 2016]



Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Sandle, T. (2015) What you need to know when volunteering for a genetic study. *Digital Journal* [online]. Available at <http://www.digitaljournal.com/life/health/volunteering-for-a-genetic-study-would-you-want-to-know/article/432932> [Accessed 16 Nov. 2016]

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What you need to know when volunteering for a genetic study

BY TIM SANDLE MAY 10, 2015 IN HEALTH LISTEN | PRINT

Would you volunteer for a genetic study? If so would you want to know if you had a rare disease? It seems that, for most people, the answer is "yes."

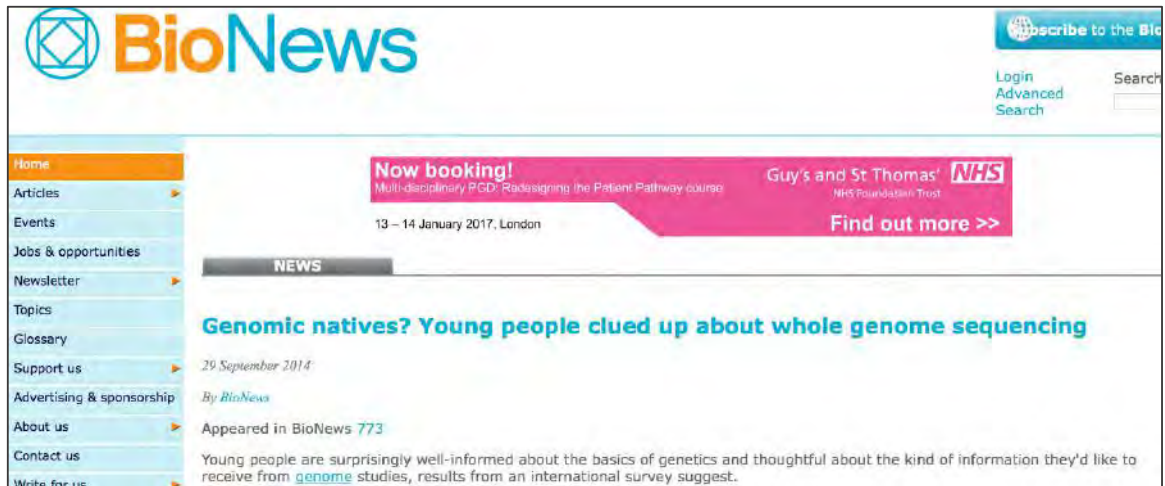
Commenting on this, the researcher who undertook the analysis, Anna Middleton of the Wellcome Trust Sanger Institute, **is quoted as saying**: "The advent of fast, efficient genetic sequencing has transformed medical research over the past decade and it's set to revolutionise clinical care in the future. Policy surrounding the use of genetic data in research and clinical settings must be directed by the views and experiences of the public, patients, clinicians, genetic health professions and genomic researchers. This study represents a first step in informing people of the issues and gathering their responses."

The outcome of the survey and its analysis has been **published in** *European Journal of Human Genetics*. The research paper is titled "Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research."

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

BioNews (2014). Genomic natives? Young people clued up about whole genome sequencing. *BioNews* [online]. Available at http://www.bionews.org.uk/page_456273.asp [Accessed on 17 Nov. 2016]



The screenshot shows the BioNews website interface. At the top left is the BioNews logo. On the right, there are links for 'Subscribe to the BioNews', 'Login', 'Advanced Search', and a search box. A navigation menu on the left lists: Home, Articles, Events, Jobs & opportunities, Newsletter, Topics, Glossary, Support us, Advertising & sponsorship, About us, Contact us, and Write for us. A pink banner for 'Now booking!' for a 'Multi-disciplinary PGD: Redesigning the Patient Pathway course' at 'Guy's and St Thomas' NHS Foundation Trust' is visible, dated 13-14 January 2017. The main article is titled 'Genomic natives? Young people clued up about whole genome sequencing', dated 29 September 2014, by BioNews. The article text begins: 'Young people are surprisingly well-informed about the basics of genetics and thoughtful about the kind of information they'd like to receive from genome studies, results from an international survey suggest.'

Middleton, A (2014). Understanding the impact of genomic data. *Wellcome Trust Blog* [online]. Available at <https://sangerinstitute.wordpress.com/2014/12/12/understanding-the-impact-of-genomic-data/> [Accessed on 17 Nov. 2016]



The screenshot shows a blog post from the Wellcome Trust Sanger Institute. The header includes the Wellcome Trust Sanger Institute logo and 'the blog inside the Institute'. Navigation links are: HOME, ABOUT, CONTRIBUTORS, TERMS AND CONDITIONS. The article title is 'Understanding the impact of genomic data', posted on 12 December 2014 by Anna Middleton. The text discusses the genomic era and the launch of 23andMe. A photo shows a family (man, woman, child) with a genetic counsellor. A caption reads: 'Genetic counsellors talk patients through implications of findings and advise them on how to share findings with family. Credits Anthea Sieveking, Wellcome Images'. On the right, there is a search box, a link to the Sanger Institute website, and a Twitter feed snippet: 'Thousands of families will benefit from pioneering programme for diagnosing rare diseases #DDD ft.com/cms /s/0/a99e16... 9 minutes ago'.

Blog and Online Journal Articles Referring to the GenomEthics Study

(section 7 of 8)

Philippidis, A. (2012) U.K. Groups Grapple with Ethical Issues Linked to Clinical Sequencing. *GEN: Genetic Engineering & Biotechnology News* [online]. Available at <http://www.genengnews.com/gen-exclusives/uk-groups-grapple-with-ethical-issues-linked-to-clinical-sequencing/77899571> [Accessed 17 Nov. 2016]



GEN Genetic Engineering & Biotechnology News SEND TO PRINTER

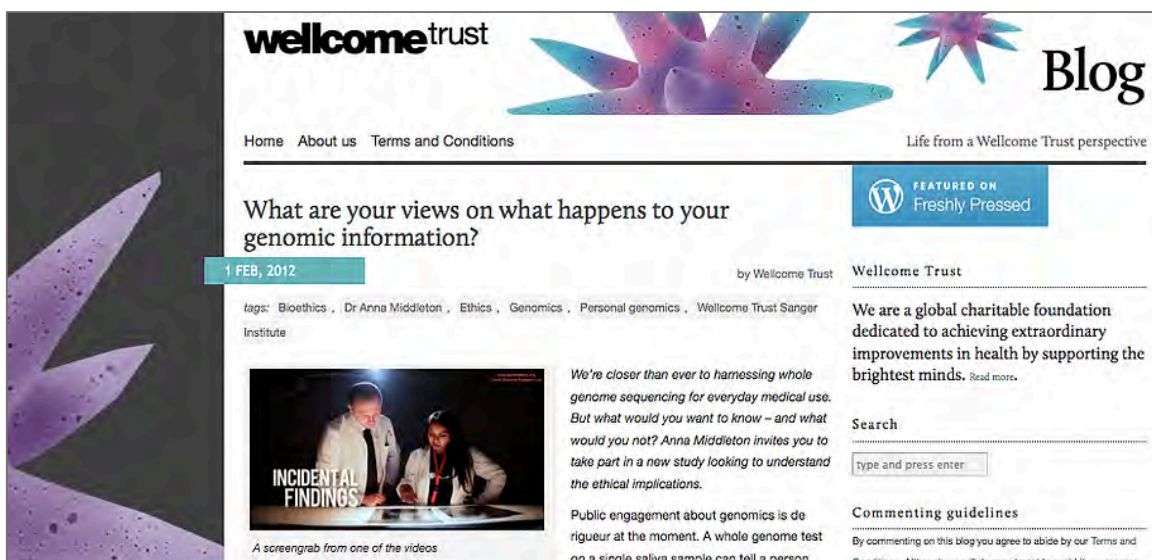
Insight & Intelligence™ : Mar 13, 2012

U.K. Groups Grapple with Ethical Issues Linked to Clinical Sequencing

Questions cover data dissemination, bioinformatics, and more education.

As a U.S. presidential panel studies the ethical issues associated with whole-genome sequencing in clinical care, it may do well to look across the Atlantic at the U.K. The Wellcome Trust Sanger Institute and research partners have a two-prong survey under way involving healthcare professionals and patients. The five-year Genome Ethics survey is designed to gather empirical data that helps guide policy decision-making on whole-genome research studies.

Middleton, A (2012). What are your views on what happens to your genomic information? *Wellcome Trust Blog* [online]. Available at <https://blog.wellcome.ac.uk/2012/02/01/what-are-your-views-on-what-happens-to-your-genomic-information/> [Accessed on 17 Nov. 2016]



wellcometrust Blog

Home About us Terms and Conditions Life from a Wellcome Trust perspective

What are your views on what happens to your genomic information?

1 FEB, 2012 by Wellcome Trust

tags: Bioethics , Dr Anna Middleton , Ethics , Genomics , Personal genomics , Wellcome Trust Sanger Institute

Wellcome Trust
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By commenting on this blog you agree to abide by our Terms and Conditions. Although we will do our utmost to avoid it, we reserve

INCIDENTAL FINDINGS
A screengrab from one of the videos

We're closer than ever to harnessing whole genome sequencing for everyday medical use. But what would you want to know – and what would you not? Anna Middleton invites you to take part in a new study looking to understand the ethical implications.

Public engagement about genomics is de rigueur at the moment. A whole genome test on a single saliva sample can tell a person

Social Media

(section 8 of 8)

9 Feb. 2015 - Invited to participate in GenomeSeqWeek at Genetic Alliance to take over the Genetic Alliance Twitter account for an hour along with Vivienne Parry OBE

GenomeSeqWeek.com

The genomic revolution is here!
Join us for our week of events exploring
how whole genome sequencing
could impact your future

TUESDAY 10 FEBRUARY

Do You Really Want to Know?

Come along to the screening of this award-winning documentary by director John Zaritsky about three families who must decide whether or not to be tested for Huntington's disease. The screening will be followed by a panel discussion chaired by Charles Sabine, a former NBC news war correspondent who has Huntington's disease.

Genesis Cinema, 93-95 Mile End Road, London E1 4UJ
6.30pm – 9pm

MONDAY 9 FEBRUARY

The future of genomics

Follow us to hear journalist and broadcaster, Vivienne Parry OBE, and bioethicist, Dr Anna Middleton, discuss the breakthroughs in genetics since the discovery of DNA over 50 years ago and answer your questions about what the future holds.

@GeneticAll_UK
7 – 8pm

Social Media

(section 8 of 8)

Facebook

A screenshot of a Facebook search for "Genomethics". The search bar at the top shows "Genomethics" with a magnifying glass icon. Below the search bar are navigation tabs: Top, Latest, People, Photos, Videos, Shop, Pages, Places. The "POSTED BY" section on the left has radio buttons for "Anyone" (selected), "You", "Your Friends", "Your Friends and Groups", and "Choose a Source...". The "TAGGED LOCATION" section has radio buttons for "Anywhere" (selected), "Cambridge, United Kingdom", and "Potomac, MD". The main content area shows a post from "Medical Law and Bioethics News" dated "January 31, 2012". The post title is "Important Wellcome Trust-backed survey on genome ethics & return of IFs" with a link to http://www.genomethics.org/about_questionnaire.html. There is a "Like Page" button. Below the text is a thumbnail image for a "Genomics Survey" and a link to "Genomethics - Home". At the bottom of the post area, it says "GENOMETHICS.ORG | BY CS16 (CLAIRE STEED)".

A screenshot of a Facebook search for "Genomethics". The search bar at the top shows "Genomethics" with a magnifying glass icon. Below the search bar are navigation tabs: Top, Latest, People, Photos, Videos, Shop, Pages, Places. The "POSTED BY" section on the left has radio buttons for "Anyone" (selected), "You", "Your Friends", "Your Friends and Groups", and "Choose a Source...". The "TAGGED LOCATION" section has radio buttons for "Anywhere" (selected), "Cambridge, United Kingdom", and "Potomac, MD". The main content area shows a post from "Medical Law and Bioethics News" dated "January 31, 2012". The post title is "Ethics and Genomic Research: 'Genomethics' « Genomes Unzipped" with a link to <http://www.genomesunzipped.org/2012/01/genomethics.php>. There is a "Like Page" button. Below the text is a thumbnail image showing two people looking at a screen with the text "MENTAL FINDINGS". At the bottom of the post area, it says "GENOMESUNZIPPED.ORG".

Social Media

(section 8 of 8)

Facebook (continued)

The screenshot shows a Facebook interface with a search bar at the top containing 'genomethics'. Below the search bar are navigation tabs: 'Top', 'Latest', 'People', 'Photos', 'Videos', 'Shop', 'Pages', 'Places', and 'G'. The main content area displays a post from 'The Family Network' shared by 'Unique - Understanding Chromosome Disorders' on March 18, 2013. The post features a photograph of a young child in a light blue shirt holding a colorful DNA double helix model. The text of the post reads: 'For those with rare chromosome disorders' followed by the photo. Below the photo, the post title 'Unique - Understanding Chromosome Disorders' is repeated, along with the date 'March 18, 2013'. A 'Like Page' button is visible to the right. The text continues: '*Your help is needed! Parents of children with genetic disorders are needed to give their voice and be heard. A survey is being done in Cambridge that gathers ...'. A 'See More' link is at the bottom of the post. On the left side of the page, there are filter menus for 'POSTED BY' (Anyone, You, Your Friends, Your Friends and Groups, Choose a Source...), 'TAGGED LOCATION' (Anywhere, Cambridge, United Kingdom, Potomac, MD, Choose a Location...), and 'DATE POSTED' (Anytime, 2016, 2015, 2014, Choose a Date...).

Social Media

(section 8 of 8)

Facebook
(continued)

Social Media

(section 8 of 8)

Facebook (continued)

Molecular Creativity



Like Message Share More

European Journal of Human Genetics - Abstract of article: Attitudes of nearly 7000 health...

The European Journal of Human Genetics is the official Journal of the European Society of Human Genetics, publishing high-quality, original research papers, short reports, News and Commentary articles and reviews in the rapidly expanding field of human genetics and genomics.

NATURE.COM

Like Comment Share

Research Ethics



Like Message Share More

Research Ethics
August 3, 2015 · €

<http://jmg.bmj.com/content/52/8/571.long>

Potential research participants support the return of raw sequence data -- Middleton et al. 52...

Health-related results that are discovered in the process of genomic research should only be returned to research participants after being clinically validated and then delivered and followed up within a health service. Returning such results may be difficult for genomic researchers who are limited...

Médicos Psiquiatras



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Médicos Psiquiatras
May 6, 2015 · €

El 98% de las personas ansia conocer los secretos de su #Genética
<http://www.nature.com/.../journal/vaop/.../full/ejhg201558a.html>

See Translation



European Journal of Human Genetics - Attitudes of nearly 7000 health professionals, genomic...

The European Journal of Human Genetics is the official Journal of the European Society of Human...

NATURE.COM

GENOMETHICS

DDD/Ethics Project

Social Media

(section 8 of 8)

Facebook (continued)

Orphanet-Italia
July 10, 2015 · 🇪🇺

UNO STUDIO SULLA PROPENSIONE A FORNIRE AI PAZIENTI I DATI DEL SEQUenziAMENTO

Un articolo pubblicato sul Journal of Medical Genetics illustra l'etica e la responsabilità dei ricercatori in merito a come fornire ai pazienti informazioni sul sequenziamento del genoma. Gli autori affermano che i risultati relativi alla salute scoperti durante un progetto di ricerca sul genoma dovrebbero essere forniti solo a chi ha partecipato allo studio, "solo dopo che questi siano stati validati da un punto di vista clinico e quindi rilasciati e ulteriormente sviluppati nell'ambito di un servizio sanitario". Tuttavia, gli stessi autori comprendono che fornire questo tipo di informazioni, potrebbe essere difficile per i ricercatori, limitati dalle risorse e dall'accesso a strumenti clinici adeguati. Gli autori analizzano la propensione di oltre 7.000 persone tra ricercatori, professionisti della genetica e professionisti del settore sanitario non genetisti chiedendo cosa farebbero con questi dati, qualora venissero forniti loro. Il risultato è stato che il 62% dei partecipanti all'indagine avrebbe avuto interesse ad utilizzarli per cercare di fornire la propria interpretazione clinica. In conclusione, sebbene gli autori non promuovano al momento la comunicazione di questi dati, soprattutto in considerazione dell'attuale incertezza interpretativa, ritengono tuttavia necessario un dibattito etico approfondito sul tema per creare un quadro etico che possa guidare la policy su come rendere disponibili i dati del sequenziamento.

Potential research participants support the return of raw sequence data. - PubMed - NCBI

J Med Genet. 2015 May 20. pii: jmedgenet-2015-103119. doi: 10.1136/jmedgenet-2015-103119....
NCBI.NLM.NIH.GOV | BY MIDDLETON A, ET AL.

Summer internship for INdigenous peoples in Genomics - SING
April 29, 2015 · 🇪🇺

British Study. 7000 people polled would like their incidental genetic findings returned to them. <http://www.nature.com/.../vaop/ncurrent/full/ejhg201558a.html...>

GENOMETHICS

DDD/Ethics Project

Social Media

(section 8 of 8)

Facebook (continued)

Joanne Loves Science
@joannelovesscience

Home
About
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Like Share Save More

Joanne Loves Science
May 14, 2015 · 🌐

Many say If researchers using their genetic data find hints of a serious preventable or treatable disease, they'd want to know.

A survey of nearly 7,000 people from 75 countries reveals that 98% would want to be informed if researchers using their genetic data stumble upon indicators of a serious preventable or treatable disease. The study, published in the European Journal of Human Genetics, offers the largest dataset, published to date, of attitudes towards issues surrounding the return of 'incidental findings' from sequencing research. Read the full open access article: <http://bit.ly/1QBx2jj> #genetics

Your DNA, your views
A survey of nearly 7000 people has revealed that 98 per cent want to be informed if researchers using their genetic data stumble upon indicators of a serious...
YOUTUBE.COM

OHSU Bioinformatics and Computational Biology

Like Message Share More

OHSU Bioinformatics and Computational Biology
April 29, 2015 · 🌐

"This finding demonstrates a disconnect between the views of those handling the findings of research and those participating in research" - Results of survey on incidental findings

European Journal of Human Genetics - Attitudes of nearly 7000 health professionals, genomic...

The European Journal of Human Genetics is the official Journal of the European Society of Human Genetics, publishing high-quality, original research papers, short reports, News and Commentary articles and reviews in the rapidly expanding field of human genetics and genomics.
NATURE.COM

GENOMETHICS

DDD/Ethics Project

Social Media

(section 8 of 8)

Facebook (continued)

The image shows a Facebook post from the page 'Nature' (@nature). The post is dated May 14, 2015. The main text of the post reads: 'A survey of nearly 7,000 people from 75 countries reveals that 98% would want to be informed if researchers using their genetic data stumble upon indicators of a serious preventable or treatable disease. The study, published in the European Journal of Human Genetics, offers the largest dataset, published to date, of attitudes towards issues surrounding the return of 'incidental findings' from sequencing research. Read the full open access article online: <http://bit.ly/1F1YLoc>'. Below the text is a video player with a thumbnail showing a laboratory setting and the title 'Your DNA, your views'. The video description says: 'A survey of nearly 7000 people has revealed that 98 per cent want to be informed if researchers using their genetic data stumble upon indicators of a serious...'. The video is from YOUTUBE.COM. The post has 48 likes, 19 shares, and 1 comment. The left sidebar shows the 'Nature' profile with navigation options: Home, About, Videos, Photos, Posts (selected), Likes, Events, and Reviews.

GENOMETHICS

DDD/Ethics Project

Appendices

1. Appendix A: Original Protocol for Ethics and Whole Genome Studies (written in 2011)
2. Appendix B: Data Protection Registration
3. Appendix C: Lone Worker Policy
4. Appendix D: Favourable REC Opinions
5. Appendix E: Creation of a Bespoke Survey

Appendices

Appendix A: Original Protocol for Ethics and Whole Genome Studies (written in 2011)

Summary of the study

The Deciphering Developmental Disorders (DDD) project uses the latest whole genome technologies to investigate 12,000 children with undiagnosed developmental delay from every Regional Genetics Service in the UK. The aim of this is to identify new genetic causes for developmental disorders. However, because the research involves looking at the whole genome (all of a person's genes), it is inevitable that clinically significant, but 'incidental', findings will be uncovered in some research participants. For example, a predisposition to developing adult-onset breast cancer may be found in a two-year-old. Such a finding is unlikely to be related to the developmental disorder and yet could still be clinically significant to the child in later life as well as to other family members. As yet there are no published large-scale studies that have gathered empirical data on views about sharing incidental findings from whole genome studies; we aim to address this.

Our objective is to ascertain the views of research participants from the DDD project, genomic researchers, genetic health professionals, laboratory staff and members of the public. Our questions focus on attitudes towards sharing incidental findings, how such findings could be categorized, what to do with findings of unknown significance, attitudes towards mining specifically for certain types of incidental findings as well as views on consenting procedures. Quantitative and qualitative research methods are used to explore the above issues.

Summary of main issues

The Deciphering Developmental Disorders (DDD) project uses the latest whole genome technologies to investigate 12,000 children with undiagnosed developmental delay and their parents from each of the 23 Regional Genetics Service in the UK. This project has already gained multi-centre Research Ethics Committee approval (number: 10/H0305/83) as well as NHS Research and Development Approval across all involved NHS sites in the UK.

The ethics/social sciences research (called 'ethics study' from here onwards) under consideration within this proposal is aligned directly to the DDD project; this additional work pertains to explore some of the ethical issues relating to the information gained from whole genome studies. REC approval has already been granted within the DDD project for contacting potential research participants to explain more about our ethics study and invite participation; we are now seeking additional REC approval to conduct the ethics study.

Appendices

All participants in the ethics study will be invited to complete an online questionnaire (and if they so choose, an interview) in their own time at home. They will not be participating on NHS premises; some of them will be recruited through the NHS (via an invitation to complete the online questionnaire at home) and REC approval has already been gathered for this initial invitation via the main DDD project. We have been advised by the sponsoring NHS R+D centre for the main DDD study that NHS Trust R+D approval is not necessary for the ethics study (letter attached) and therefore we will not be seeking this.

The aim of the DDD project is to identify new genetic causes for developmental disorders. However, in doing so, clinically significant 'incidental findings' may be uncovered. For example, a genetic predisposition to developing adult-onset breast cancer may be found in a two-year-old. Such a finding is unlikely to be related to the developmental disorder and yet could still be clinically significant to the child in later life as well as to other family members. Incidental findings could include variants of known and unknown significance, information about life-threatening and serious conditions and information about carrier status for a whole variety of genetic conditions. In the DDD research project, incidental findings will not be revealed to participants until more is known about the ethical implications of reporting such results. However, pressure is mounting from policy makers and ethicists to share clinically significant incidental findings, the thinking being that it is unethical to withhold genetic information that could enable the research participant to take preventative or therapeutic action to protect their health. Whilst there is often sympathy with this position, some genomic researchers are concerned that the time spent searching for, interpreting and reporting incidental findings unrelated to the research aims might jeopardise attaining those aims.

As yet there are no published large-scale studies that have gathered empirical data on any of these ethical issues; we aim to address this omission as part of this ethics study. We are ascertaining the views of research participants from the DDD study, genomic researchers, health professionals, laboratory staff and members of the public. The questionnaire will be freely available online and thus has the capacity to be viewed widely and also by people who have not received a direct invitation from us.

The study questions focus on attitudes towards sharing incidental findings, how such findings could be categorized, what to do with findings of unknown significance, attitudes towards mining specifically for certain types of incidental findings as well as views on how consenting procedures in whole genome studies should be structured. Our ethics/social sciences study uses a mixed methods approach, utilising both quantitative and qualitative techniques. Throughout 2011-2012 more than 25,000 people will be invited to participate in the online

Appendices

questionnaire and from these 50-100 people will be invited for a face-to-face interview.

The principal research question/objective

- This is an exploratory study; the objectives are to investigate the following:
- Attitudes towards sharing of 'pertinent findings' from whole genome studies
- Attitudes towards sharing of 'incidental findings' from whole genome studies
- Attitudes towards receiving information relating to different categories of genetic condition
- Attitudes towards the sharing of raw genomic data
- Attitudes towards genomic researchers having a duty to search for incidental findings
- Attitudes towards having a 'gatekeeper' of genomic data
- Attitudes towards possible consenting procedures for genomic studies

Scientific justification for the research

Exploratory whole genome studies involve searching through all of an individual's genes looking for variants in similarly affected people that could contribute towards causing a particular clinical phenotype ('phenotype' - is the set of clinical features a person might have, e.g. breast cancer). Whole genome studies, by virtue of involving all 20,000+ genes, inevitably produce large volumes of genetic data. Some of this may be directly linked to the phenotypes under study (considered 'pertinent findings') and others may be completely unrelated (considered 'incidental findings'). There is no universally accepted definition of what an incidental finding is (Wolf, Lawrenz et al. 2008), and broadly speaking this could include variants of known and unknown clinical significance, variants linked to highly penetrant, serious, life-threatening conditions, non-paternity or ancestry data.

There is evidence to suggest that research participants in genetics studies want to receive pertinent findings relating to the medical condition under study (Wendler and Emanuel 2002). However, little is known about what research participants think about incidental findings, including clinically significant information relating to medical conditions *unrelated* to the medical condition under study. There is much discussion in the medical, ethics, genetics and social sciences literature about the merits and pitfalls of sharing genomic information in a research and clinical setting (Kohane, Masys et al. 2006; Renegar, Webster et al. 2006; Miller, Giacomini et al. 2008; Knoppers and Laberge 2009) and increasing support for the position that it is ethical to share incidental findings from whole genome studies (Knoppers, Joly et al. 2006; Wolf, Lawrenz et al. 2008; Beskow and Burke 2010; McGuire and Lupski 2010). 'Even pure scientists can and should advance research

Appendices

subjects well-being and respect their autonomy by making appropriate disclosures of potentially significant incidental findings' (Miller, Mello et al. 2008).

Empirical data on the attitudes, values and beliefs of research participants in receiving genomic results is limited. There have been a few small-scale qualitative interview studies (Miller, Giacomini et al. 2008; Miller, Hayeems et al. 2010) and each has emphasised the need for further research in this area. To our knowledge there are no large-scale quantitative studies that clearly demonstrate attitudes towards sharing of incidental findings. Our ethics study proposes to address this.

Design and methodology

Online Questionnaire

In preparation for obtaining REC approval, the study questionnaire design process has been started, this is so that the REC committee can see a copy of the preliminary questionnaire and thus will have full information about the subject matter covered. As with any questionnaire design process, extensive background work is required to create the questionnaire. A transparent summary of this work is included.

A nonstandard, quantitative questionnaire will be used in this study, including 24 closed questions. The questionnaire has been created using a systematic approach that adheres to robust principles of questionnaire design (Denscombe 2005; Aday 2006; Lietz 2010; Vicente and Reis 2010). See Appendix A for details of the questionnaire creation process, including piloting, face validity and reliability testing.

An informal systematic review of the literature has been completed. The databases PubMed and Scopus were used, with the search terms 'genomic, incidental finding, research study, whole genome study, GWAS, ethics, results sharing, data sharing'. From this literature review, broad themes were drawn that formed the basis of the questionnaire. These themes addressed two needs: 1) they covered issues that other researchers had identified as important for further study 2) they covered issues that policy makers had anecdotally identified as important for practice, but for which there was no empirical data to support them.

The selected themes were checked for face validity with internal and external stakeholders in the research. As a result of these discussions a preliminary set of potential questions were created that related to each theme. These questions were debated and discussed in an informal focus group with 6 practicing genetic counsellors. Genetic counsellors were chosen because they are health professionals directly involved in recruitment into genetic and genomic research studies but also they work directly with members of the public. They also have a wealth of experience in genetics. Thus, this group were knowledgeable about

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how complex issues surrounding genetics (i.e. ethical implications of genomic research studies) can be translated into lay language for the public – of key importance to the study questionnaire. The focus group offered feedback to the research team on whether the proposed questionnaire themes and types of questions appeared suitable and whether they were acceptable for gathering relevant and useful data.

The questionnaire structure has encompassed best, evidenced-based practice for questionnaire design. For example, questions are short (Fink 2003) and have less than 20 words per sentence (Oppenheim 1992), closed-end responses are listed vertically (Aday 2006) and demographic questions are positioned at the end (Lietz 2010). However, as our questionnaire is to be delivered online rather than via post, we have had to adhere to additional, evidence-based practice for online questionnaire design.

'Although some of the design principles established for mail surveys may be translated to web surveys because both methods are self-administered, others require specific treatment in the Internet context because the technical features of the Internet allow a respondent-survey interaction that is distinct from that of paper questionnaires (Couper, Traugott et al. 2001)' p252 (Vicente and Reis 2010)

Thus, we have included the use of video to deliver the information required to answer each question. This has the advantage over written text that would only be suitable to a postal questionnaire in that it is visually engaging, interesting and creative. Each mini-film that accompanies the questions will be deliberately short (less than one minute), so that it is not time-consuming to watch and will use a mixture of video footage, animation and voice-over to relay the intended messages (see later for details).

In accordance with 'good' practice for web-based questionnaire design, in-depth consideration has been given to the online style of the questionnaire (Vicente and Reis 2010). For example, there is evidence to suggest that participants in an online questionnaire make an initial assessment of how many questions there are and how long they perceive it will take them to complete them, if they can't immediately make this assessment then they are not motivated to continue and may decline participation (Ganassali 2008). Thus our online questionnaire has been deliberately formatted so that it is easy to see how many sections there are to the questionnaire, and particular attention has been paid to making the navigation experience easy so that participants can easily see how much progress had been made as they are working through the questions (Vicente and Reis 2010). In addition to this there is evidence to suggest that there is less of a dropout rate and question omission rate if the questions are presented individually and the research participant has to actively click a button to reveal the next question as opposed to needing to scroll down the screen (Lozar Manfreda, Batagelj et al. 2002).

Appendices

Therefore, we have formatted our questions using multiple pages, so that participants don't have to scroll down lots of text in order to answer the questionnaire. The longer the online questionnaire the higher the dropout rate (Ganassali 2008), thus our questionnaire has been designed to only take approximately 15 minutes to complete (considered 'short' in terms of online questionnaires). Online questionnaires that have a plain background have a lower dropout rate and higher completion rate than those with bright colours (e.g. purple and pink) (Dillman, Tortora et al. 1998), therefore we have chosen a plain dark background with light text - this configuration is thought to support best practice formatting for visually impaired research participants. The questionnaire has also been checked to ensure that it is appropriate for an International audience - UK-centric words such as 'GP' have been explained via a definition in the glossary and the socio-demographic data will contain variables that are applicable to any participant, irrespective of geography.

The questionnaire is being interrogated via 5 systematic pilot studies. Three of the pilot studies deliberately involve participants of varying ages, ethnic backgrounds and professional experiences; they will also involve representative people from the three target groups (lay members of the public, health professionals and genomic researchers). Between each pilot test further face validity testing will be done with internal and external stakeholders (nationally and internationally), including the involvement of a statistician, to check that the questions still make sense and appear to adequately measure the issues of interest. Two of the pilot studies will be conducted specifically to test the reliability of the questions. Participants in these studies will be from the three groups (lay public, health professionals and genomic researchers) and questionnaires will be completed at two different time points, 2 months apart. This is to check that the questions ascertain the same results over time (and thus are likely to be reliable measures).

From preliminary work done on the questionnaire validation process the following themes have been identified and will form the basis of the questionnaire content:

- Sharing of pertinent findings (should pertinent findings from whole genome studies be shared with research participants?)
- Sharing of incidental findings (should incidental findings from whole genome studies be shared with research participants?)
- Categorizing of incidental findings (what categories of incidental findings are research participants interested in knowing about?)
- What to do with raw genomic data (should raw genomic data be shared with research participants, what would they do with this?)
- Duty of genomic researchers (should genomic researchers actively mine for incidental findings or should they only consider them if they arise while they are searching for pertinent findings?)

Appendices

- Delivery of genomic information (should someone filter genomic results? If so, who?)
- Consent for genomic research (is it acceptable to have a 'flexible consent' process?)

Each of the above themes considers complex ideas of what a genome is and requires some level of understanding about genetics. Thus, particularly for the lay members of the public who will be participating in the project, there is great importance that these themes are carefully explained in lay language. As the questionnaire will be available online, video will be used to describe the required concepts in an engaging and interesting manner. Two independent film makers have been consulted and have assisted in the creation of 7 scripts that will be turned into video that will appear in each of the 7 themes in the questionnaire. As the creation of the film is expensive this will not be done until the questionnaire has gone through the REC approval process, this is just in case the REC committee wish to make changes to the questionnaire.

Semi-structured Interview

Participants who have completed the online questionnaire will be invited to participate in an interview, if they so choose. If they are interested they can leave their contact details and a member of the ethics research team will contact them to arrange a date to interview them at a place of their choice (e.g. their home or work). Interviews will be conducted in the UK only. From those who choose to leave their contact details, participants will be selected for interview depending on their responses to the questionnaire. The aim being to select as diverse a group as possible, including 'deviant cases', (Atkinson, Coffey et al. 2003) so that there is a real spread of views about the 7 themes above. Participants will also be from the three stakeholder groups: lay public, health professionals and genomic researchers.

Involvement of patients, users, members of the public:

Beverly Searle CEO of 'Unique' the support group for families and individuals affected by rare chromosome disorders is on the Scientific Advisory Board for the DDD project. She has been directly involved in the ethics questionnaire design process and members of Unique (parents of children with diagnosed and undiagnosed developmental disorders) have participated in the piloting of the ethics questionnaire. Thus, as patients and users of genome research studies (and also as members of the public) they have contributed directly to ensure the ethics questionnaire is appropriate and sensitive for members of the public.

Appendices

Principal inclusion criteria

We propose to include 3 broad groups of participants who will be recruited in the online questionnaire and interview study:

lay members of the public
health professionals
genomic researchers.

These broad groups can be further refined into:

Primary stakeholders (parents of children involved in DDD; genetic health professionals, including clinical lab staff; genomic researchers)

Secondary stakeholders (research participants in genomic studies (non-DDD) plus people totally unconnected to research and/or genetics; health professionals unconnected to genetics/DDD)

The principal inclusion criteria is that research participants fall into the above groups. Participants have to be over the age of 18. They can live anywhere in the World but to participate in the interview study they need to be available to meet in the UK. Anyone who understands written and spoken English can participate; the online questionnaire will be available with subtitles for hard of hearing users but will not be translated into other languages.

Principal exclusion criteria

The principal exclusion criteria is anyone who doesn't have access to the Internet.

According to data from the 2011 census from the Office of National Statistics, 82.5% of the British population have access to the Internet and 60% (30.1 million) adults in the UK access the Internet every day or almost every day (www.statistics.gov.uk/cci/nugget.asp?id=8).

Internet use is directly linked to age; people aged 65 or older are least likely to use the Internet whereas 99% of 16-24 year olds regularly use the Internet. Participants in whole genome studies have a variety of ages, but anecdotally, the vast majority are under the age of 65. Thus, we feel that although using the Internet as the method for involvement in the ethics study will preclude some people from participating, it is likely that this group are the least interested in (nor appropriate for) whole genome studies anyway.

Sample size

20,000 research participants and health professionals involved in the DDD project will be made aware of the online questionnaire. 5,000 genetic health professionals and genomic researchers will be contacted through professional email list-serves. If we anticipate a 20% response rate as predicted for good quality online

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questionnaires (Vicente and Reis 2010) then we predict our sample size to be approximately 5,000.

Of these, between 50-100 will be invited to have an interview. They will be selected on the basis of their answers to the online questionnaire as well as their willingness to be interviewed.

How long do you expect each participant to be in the study in total?

Participation in the online questionnaire takes 10 minutes. If participants volunteer to have an interview, this will last approximately one hour. Recruitment into the ethics study will last 2 years and the study as a whole lasts 5 years.

What are the potential risks and burdens for research participants and how will you minimise them?

Online questionnaire

The burden for research participant is taking the time to complete the online questionnaire. We have tried to minimise the inconvenience of this by making the questionnaire as visually interesting and as rewarding as possible, with the provision of video delivered in a creative and fun manner. We have also limited the questionnaire in size so that it should only take 10 minutes to complete and thus not be too arduous in terms of time. There are limited risks from completing the questionnaire – participation is anonymous (unless participants choose to leave their contact details because they would like to have an interview). For those participants who do leave their contact details these will be stored on a secure network and subject to Wellcome Trust Sanger Institute data protection policies.

Assessment of risk: low

Interview

Interviews will be conducted with people who volunteer themselves (i.e. they show an interest and are motivated); this recruitment mechanism was chosen so that it is the least burdensome for participants. The subject matter to be discussed in the interview is neither sensitive nor embarrassing; moreover, the interview will involve a discussion about sharing of data from whole genome studies and will expand on themes already introduced in the online questionnaire. The only burden associated with the interviews is the hour of participant's time required to do the interview, plus the time taken to organise this.

Assessment of risk: low

The ethics study is being conducted independently of the DDD molecular study, thus neither questionnaires nor interview data will be linked directly to genomic data.

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What is the potential benefit to research participants?

The potential benefit to research participants is a gain in knowledge about whole genome studies and the opportunity to express their views about what should count as good practice in this important area of research practice.

What are the potential risks for the researchers themselves?

The potential risks to the researchers are very low. The only risks relate to visiting research participants in their own homes to conduct the interviews. The researcher doing the interviews is experienced in home-visiting in both a clinical and research context.

We will follow best practice for 'Lone Workers Visiting Research Participants', e.g. as recommended by Faculty of Health Sciences at the University of Dublin and Keele University. These recommendations suggest a researcher visiting a research participant in their home should: carry an official identity card, carry a mobile phone, maintain a visit proforma so that it is easy for colleagues to see exactly where the researcher has gone, have car breakdown cover etc. Our Lone Worker Recommendations are attached.

How will potential participants, records or sample be identified? Who will carry this out and what resources will be used?

No NHS patient records are involved in the ethics study. There is also no link to the molecular arm of the DDD project, thus participants in the ethics study are not linked directly to any samples or information they may have provided separately to the molecular DDD team or to their local health professional.

Participants completing the online questionnaire will be randomly assigned an identifying number; this number will be used to identify their interview if they choose to have one. The ethics researcher (Dr Anna Middleton) will hold the key that links the identifying number to the research participant (and their personal information if they chose to leave this). All participation is anonymous unless participants choose to leave their contact details. They only need to do this if they want to be involved in an interview or want to receive results of the study. It is anticipated (based on previous studies of a similar nature) that approximately 20% of participants completing the online questionnaire will leave their contact details.

Participants having an interview will have the choice of being video or audio-taped. The recordings will be listened to by a member of the research team and the words will be transcribed into written text. It is this written text that will be analysed for the research study. There will be no identifying information, such as a name or address on this written transcript so for most people reading this text, they will not be able to link this back to the person who gave the interview. Written

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quotes from the transcript may be used in any reports or papers that are produced from the research, but again these will not contain any identifying information that relate to the research participant.

If the research participant chooses to be video-recorded then there are a number of options available to them in terms of how their interviews can be used. They could consent to just having an anonymous written transcript produced from the interview and nothing else. Or they could consent to clips from their video being used in the dissemination phase of the study. When the research is finished members of the research team will present this work at conferences and to a public audience, if they consent, then small clips from the research participant's interview could be shared in such an arena, to demonstrate particular opinions. Thus the visual image from the research participant would be shared and their identity in this sense would be accessible (but not their name or address). The research participant could also consent to these clips being shown on the Internet. Research participants will be able to see their video first before they decide finally on any of these choices.


The consent form for the interviews will also ask the research participant to consider giving permission for other researchers in the future to analyse their interview. This could take the form of looking at the written transcript or watching the original video. Any future and different research projects would require additional ethical approval from a research ethics committee.

All video and audio data files will be stored electronically on the Wellcome Trust Sanger Institute secure network. All online questionnaires will be stored, with encrypted backup, served via https. Thus the computer storage of data is very secure.

Will any participants be recruited by publicity through posters, leaflets, adverts or websites?

The research team are currently displaying the following information on the www.ddduk.org website to describe the ethics study:

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Ethics research

- What information would you want to know about your own genome?
- Should it be the responsibility of medical researchers to provide any information that people might want to know about their genome?
- Are there medical conditions that you would like to know about from your genome that are unrelated to developmental disorders?

Ethics research is a core part of the DDD study. We hope that it will pave the way for an informed public debate about the ethical and social implications of the use of whole genome sequencing in health care and its appropriate control.

What do you think?

The DDD research team is keen to gather your views on the ethical implications of whole genome testing. **We are currently in the process of developing an online questionnaire, so please visit again soon to give us your views.** Anyone can answer the questionnaire whether they are involved in the study as a participant or researcher or are just an interested member of the public. Your views are important in shaping how these new technologies are applied in medicine and we hope you will want to take a few minutes to share your thoughts.



Key questions

One of the major questions about whole genome technologies is what to do with findings that don't relate to diagnosis of the disorder being studied. Because of the enormous variation that exists between all of our genomes, everyone will have some of these 'incidental' findings. Although we don't know enough yet to understand what most of these findings mean, some could be important for your health or family relationships. For example, a strong genetic predisposition to Alzheimer's disease is irrelevant to the diagnosis of a child's developmental disorder, but may be relevant to their future health or the health of their family. Researchers and clinicians are currently struggling to work out how to deal with these findings in a responsible and ethical way. We aim to find out how to deal with such issues by gathering opinions from the public, health care professionals and genomic researchers.

This site is hosted by the Wellcome Trust Sanger Institute. Terms and Conditions.

The above will form the basis of any publicity material that is advertised about the ethics study. However, it should be noted that as the questionnaire for the study is available freely on the Internet the researchers have no control how others choose to advertise the study (e.g. charities and support groups for people who take part in genomic research studies may choose to encourage their members to participate in the study and how they do this is up to them).

How and by whom will potential participants first be approached?

As participants in the study are recruited through an open access online questionnaire that is available to anyone to complete, it is likely that this will be propagated virally (i.e. people see the questionnaire, enjoy filling it in and tell their online friends about it). Thus the researchers have no control over how far and wide the questionnaire will be distributed, nor specifically, how the questionnaire will be advertised by others.

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Parents invited to participate in the molecular studies involved in the DDD project will be given the details of the website for the online questionnaire. This initial contact, made by the genetic health professionals from each of the 23 regional clinical genetics services, has already received REC approval when the molecular DDD project was assessed (ref: 10/H0305/83). The genetic health professional will provide the details of the website together with the information they provide about the molecular studies. Parents can then choose if they wish to visit the website with the online questionnaire or not. They will not be chased up if they choose not to pursue this.

Genetic health professionals who are recruiting research participants in the molecular arm of the DDD project will be provided with the website address for the online questionnaire. This will be sent to them in an email and also will be available in their monthly newsletter. Genetic health professionals can then choose to complete the online questionnaire if they wish, in their own time at home.

Members of the charity Unique will be approached via the Unique Facebook page. Beverly Searle, CEO of Unique, the charity that supports parents and individuals with chromosome disorders is on the Scientific Advisory Body for the DDD project and is willing to support recruitment into the ethics study. Beverly will put up a notice on their Facebook page with a link to the ethics questionnaire.

Email list servers for genetic health professionals and genomic researchers will be used to advertise the link to the online questionnaire. For example, the Association of Genetic Nurses and Counsellors (UK) and National Society of Genetic Counsellors (US) and Wellcome Trust Sanger Institute and European Bioinformatics Institute will be targeted.

Informed consent from or on behalf of research participants

For completion of the online questionnaire consent is deemed implicit if research participants choose to fill it in. They are free to ignore it and thus decline participation.

For participation in the interviews informed consent will be obtained.

Informed consent in writing?

For participation in the interview section of the ethics study, participants will initially volunteer themselves for an interview by leaving their contact details at the end of the online questionnaire. They will then be contacted by a member of the research team and sent a copy of the Information Booklet and Consent Form (both attached), a provisional appointment will then be booked for the interview. Participants will be offered the choice of being audio or video-recorded and they

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do not need to decide for definite until the day of the interview (the researcher will bring equipment for both). Participants will be given at least a week before the initial contact and the interview date and in this time they are expected to read the Information Booklet and Consent Form. When the researcher arrives to do the interview she will go through the Information Booklet and Consent Form in more detail and take written consent before the interview commences. If the research participant is happy to be video-recorded then they will be sent an electronic copy of the interview (after this has been completed) so they can view this. If on seeing their interview, they wish to change any of the video sections of the consent form then they can do this by letting the research team know within one month of the interview.

How long will potential participants have to decide whether or not to take part?

After the initial contact has been made and the Information Booklet and Consent Form have been sent, the research participant has at least a week to decide if they wish to proceed with the interview.

If research participants choose for their interview to be video-taped then they will receive a copy of this on CD/DVD to keep and they can check this to make sure they are happy with everything they said in the interview. A month after receiving this, if the research team haven't heard from the research participant we will assume that the research participant is content for us to use this video in the ways they consented. If they wish to withdraw from the study they need to do this within one month of the interview, and can do this by contacting the ethics study team. Then all data will be destroyed, e.g. consent form, interview tape. After this time the anonymous written transcript will have been involved in an aggregate analysis and so it will not be possible for us to withdraw and destroy this.

If research participants consent to clips from their video being shared publically within presentations delivered as part of the dissemination phase but then later change your mind on this (e.g. after 1 month) then they need to let us know immediately. It may be possible from that point onwards to withdraw individual video clips from the dissemination.

What arrangements have been made for persons who might not adequately understand verbal explanations or written information given in English, or who have special communication needs?

The online questionnaire is in written English and the video is provided with a voice over and subtitles. The subtitles are particularly focussed at research participants who are hard of hearing; the online questionnaire has been written (and formatted) in such a way to enhance the readability for research participants who are visually impaired (dark background, light text, increased font size).

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Due to the extensive cost involved, as well as the difficulty in translating complex genetic language into other languages (and thus the need to bring in external experts in genetics and non-British languages) (Shaw and Ahmed 2004) we have decided to make no arrangements to translate the questionnaire into any other languages.

What steps will be taken if a participant, who has given informed consent, loses capacity to consent during the study?

If a research participant loses capacity after they have completed the online questionnaire then this questionnaire will still be included in the study. There is no mechanism in place to learn if a research participant has lost capacity and indeed the vast majority of the questionnaires will be anonymous.

If a research participant loses capacity after they have completed the interview, and the research team are informed of this, then the interview and any identifying information will be destroyed if requested by the research participant's guardian.

How will the confidentiality of personal data be ensured?

The only personal data that will be stored are the names, email addresses and postal addresses of participants who volunteer to be interviewed. This information will be stored electronically, within encrypted backup, on the Wellcome Trust Sanger Institute computer network. The access to this is restricted and ring fenced so that it complies with Level 4 category data storage of the WTSI Human Genetics Data Security Policy (Feb 2011), attached.

In the interview phase of the study a written transcript of the interview will be stored, together with a randomly assigned code. The transcripts are what will be analysed by the research team and these will be anonymised, thus no identifying information will be contained within them.

The video data (if research participant's consent to their interview being filmed) will be stored electronically on the same access-restricted, ring-fenced network as provided above, abiding by the WTSI Human Genetics Data Security Policy (Feb 2011).

Who will have access to participants' personal data during the study?

The only people to have access to the participants' name and address (that have been voluntarily supplied by the research participant) are the researcher(s) who will organise and complete the interview. These same researchers will also have access to the names and addresses of people who want to receive the study results. Once the interviews have been completed and the study results have been sent, then all names and addresses will be destroyed.

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Storage and safeguarding data

All data will be stored according to intensively secure practices (for Level 4) as detailed in the WTSI Human Genetics Data Security Policy (February 2011) (policy attached). This policy is intended to protect highly personal genomic data but can also be applied to the questionnaire and interview data as well as the voluntarily submitted participant names and addresses collated in our ethics study. In order to comply with the Data Protection Act, the ethics study also has a nominated Data Controller who is registered with the Information Commissioner's Office. In addition to this the ethics study will be registered with the HMDMC (Human Materials and Data Management Committee at the WTSI), this registration requires the demonstration of best practice to safeguard and store research and personal data and through this regular checks will be made to confirm this is happening in practice. The paperwork for the HMDMC will be completed in Autumn 2011 once the infrastructure for this has been created by the Policy writers at WTSI.

The signed consent form for the interview (containing research participants name and signature, but no other identifying information) will be collected by the interviewer at their visit. It will then be stored in a locked cabinet at the Wellcome Trust Sanger Institute. This will be kept for the duration of the research project and destroyed at the end of this. However, if the research participant consents to their interview data being involved in future research involving different researchers (who will also need to obtain Research Ethics Approval from an ethics committee for a different use of this data) then their consent form will be handed over with the interview data. This is so that the next researchers know that consent has been provided for them to access this data for use in future research.

If a research participant chooses to provide their contact information so that an interview can be arranged, e.g. address, telephone and email address, then this will be stored on the secure network (backup encrypted, ring-fenced, restricted-access) at the Wellcome Trust Sanger Institute. All of this personal information will be destroyed at the end of the research project and will not be shared with anyone outside of the research team. Even if consent is provided for the interview data to be involved in future research any personal contact details will not be forwarded on to the next research team (the only information they will see is a name and signature on the consent form).

If the research participant consents for their interview to be audio-taped then after this has been transcribed into written text the original audio-tape will be destroyed. If the research participant consents to be video-taped but only so that a written transcript can be created for analysis (and thus no video is used in the dissemination phase of the study), then the video-tape/file will be destroyed as soon as the written transcript has been created. The written transcript will be

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coded so that it is not possible to identify the research participant. Anything that the research participant chooses to say within the body of the interview, such as their address or name, will be deleted from the written transcript. Thus all written transcripts will be completely anonymous.

If the research participant chooses for their interview to be video-taped and also consents for clips from the video to be used in the dissemination phase of the project then, between working on it, this will be stored on a CD/DVD in a secure, locked filing cabinet at the Wellcome Trust Sanger Institute. We will also store this on the computer network at the Wellcome Trust Sanger network (backup encrypted, ring-fenced, restricted-access). It is worth mentioning that the computer network is fastidiously maintained and has very high levels of security against external hackers.

Will the research be registered on a public database?

The DDD project (including the ethics arm of this) will be registered on the NIHR Clinical Research Network Portfolio database.

How has the scientific quality of the research been assessed?

Independent external review
Review within the research team

The DDD research proposal, including the ethics study, was sent to several independent external reviewers appointed by the Health Innovation Challenge Fund administration.

What is the primary outcome measure for the study?

The primary outcome measures are to gather the following:
Attitudes towards sharing of pertinent findings from whole genome studies
Attitudes towards sharing of incidental findings from whole genome studies
Attitudes towards receiving information relating to different categories of genetic condition
Attitudes towards the sharing of raw genomic data
Attitudes towards genomic researchers having a duty to search for incidental findings
Attitudes towards having a 'gatekeeper' of genomic data
Attitudes towards possible consenting procedures for genomic studies

Please describe the methods of analysis (statistical or other appropriate methods, e.g. for qualitative research) by which the data will be evaluated to meet the study objectives.

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Descriptive statistics will be used to explore the quantitative questionnaire data, using cross-tabulations, chi squared analysis and logistic regression. A full time statistician will work on the data.

The semi-structured interviews will explore in more depth some of the issues introduced in the online questionnaire. The interviews will be transcribed into written text and analysed iteratively, thus analysis will begin before all the interviews have been completed (Coffey and Atkinson 1996; Atkinson, Coffey et al. 2003). Interviews will be conducted until there is a saturation of themes (Flick 2006). A constant comparative, thematic analysis will be applied (Silverman 2006), starting with an open coding procedure (Strauss and Corbin 1998) and then refining with axial coding (Flick 2006). Participants can provide open, free-text comments in some sections of the online questionnaire. These too will be subjected to a thematic analysis as above.

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Appendix B: Data Protection Registration



HMDMC Data Protection Registration Version 2.0 031011

[WTSI HMDMC Data Protection Register Number](#)
(For Office Use Only)

DP11/001 (Update 2)

Wellcome Trust Sanger Institute Data Protection Registration

This form should be used to apply to the WTSI Human Materials and Data Management Committee (HMDMC) for inclusion of a research project on the WTSI Data Protection Register. All projects storing data at Level 4 (personal data) of the WTSI Human Genetics Data Security Policy* need to be registered with the HMDMC and the WTSI Data Controller. This application should be made in addition to any HMDMC approval obtained for the use of human samples (where relevant).

If the data has been collected from an organisation outside of the WTSI, then the Data Subject must have given their consent to both the collection of the data and the transfer of the data to the WTSI.

Please send the completed form to Carol Smees, Regulatory Adviser (cs14@sanger.ac.uk).

1	Name of WTSI Group Leader	Matt Hurles
2	Name of Data User(s) at the WTSI	Anna Middleton
3	Name of Data Collector(s) and their Institution <i>(Note: This is the person who <u>originally</u> collected the personal data.)</i>	Anna Middleton, WTSI
4	Name of Data Controller, e.g., Genome Research Ltd.	Genome Research Ltd.
5	Name of Sanger Institute project(s) in which the data is/are to be used. If applicable, please ensure that the project title clearly relates to the study title entered in <i>Sequencescape</i> (the WTSI DNA pipelines tracking system).	Exploring the Ethical Implications of Whole Genome Studies (part of the DDD project)
6	Brief description of the project(s) in which the personal data are to be used.	The Deciphering Developmental Disorders (DDD) project uses the latest whole genome technologies to investigate 12,000 children with undiagnosed developmental delay from every Regional Genetics Service in the UK. The aim of this is to identify new genetic causes for developmental disorders. However, because the research involves looking at the whole genome, it is inevitable that clinically significant, but 'incidental', findings will be uncovered in some research participants. For example, a predisposition to developing adult-onset breast cancer may be found in a two year old. Such a finding is unlikely to be related to the developmental disorder and yet could still be clinically significant to the child in later life as well as to other family members. As yet there are no

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	<p>published large-scale studies that have gathered empirical data on views about sharing incidental findings from whole genome studies; we aim to address this.</p> <p>Our objective is to ascertain the views of research participants from the DDD project, genomic researchers, genetic health professionals, laboratory staff and members of the public. Our questions focus on attitudes towards sharing incidental findings, how such findings could be categorized, what to do with findings of unknown significance, attitudes towards mining specifically for certain types of incidental findings as well as views on consenting procedures. Quantitative and qualitative research methods are used to explore the above issues.</p>	
7	Name(s) of Research Ethics Committee(s) (REC) approving project	Cambridge South
8	Date(s) of REC approval(s)	26 th August 2011
9	REC approval reference number(s) and, where relevant, HMDMC approval number(s)	11/EE/0313
10	Who are the Data Subjects, e.g., patients, families of patients, controls, employees, healthy volunteers, clinicians?	<p>Healthy volunteers:</p> <ol style="list-style-type: none"> 1. lay members of the public (including parents involved in the molecular arm of the DDD project) 2. genetic health professionals 3. genomic researchers
11	Are the Data Subjects adults, children, or adults lacking capacity?	Adults (no children and no adults lacking capacity)
12	Where will the data be stored, e.g., encrypted laptop, non-networked UNIX file system? Please refer to the Human Genetics Data Security Policy for further information (https://helix.wtqc.org/services/human-genetics-data-security-policy)	<p>All personally identifiable data (such as postcode, email address, name and address, when provided) will be stored at Level 4 according to the WTSI Human Genetics Data Security Policy, i.e. data will be stored on a restricted access, ring-fenced, backup encrypted network. All other research data (anonymous questionnaire answers and video interview data) will be classed as and stored at Level 3. The video interview data will not contain any personally identifiable information (e.g. name/address etc) other than the visual image of the participant. David Davison and Tim Hubbard have agreed</p>

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	that this classification of Level 3 is acceptable (1 st Feb 2012)
13 How will the data be stored, e.g., electronic database, videos, avi files, paper files?	<p>This is a social sciences study. The 'data' are attitudinal answers to an online questionnaire and interview.</p> <p>The data takes the form of written transcripts, audio/video (avi) files and hard copies of written consent forms. None of the data contains sensitive, personal or embarrassing information (including no data in the list provided at the end of this form). However, if they so choose, research participants can leave their email address if they would like to receive aggregate research results or their name and postal address if they would like to be interviewed. It is this latter information that is considered 'personal' and subject to rigorous protection issues.</p> <p>Research participants are being asked to volunteer their views about sharing incidental findings from whole genome studies. No biological or phenotypic data is being gathered.</p> <p>Electronic data will be stored, with restricted access, it is also ring-fenced and has encrypted backup. The vast majority of online questionnaires will have been completed anonymously. If participants choose, they can leave their contact details so that they can receive study results or so that they can volunteer to be interviewed. It is this personally identifiable information that requires the upmost electronic protection. This personal data will only be stored for a finite period of time. Once the study results have been emailed or an interview has been arranged and completed then at that point the contact details will be deleted. There will be no paper storage of this data.</p> <p>If participants volunteer to be interviewed then they will be recorded (audio or video – their preference). If they choose an audio recording then this will be destroyed after this has been listened to and a written transcript has been created from this. The written</p>

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transcripts will contain no identifiable information and thus will be anonymous. These will be stored electronically. If participants choose to be video-recorded they can consent to various uses of this. If they select for their video (and thus a visual image of themselves that reveals their identity) to be used in the dissemination phase of the study then small clips from their video will be aired publically. They can also consent to these being used on the Internet. Whilst the video is being worked on it will be stored electronically at Level 3. There will be no hard copies of the video.

With regards to the video recording; the filming will take place on a digital camera where footage is saved as an avi file (i.e., no tapes or discs used). The avi file will then be downloaded and electronically stored according to the Level 3 Data Security Policy. There will be no names and addresses attached to the file (so no personal data in that sense) but obviously there will be a visual image of the research participant and they will have consented to this being looked at and stored.

If volunteers choose to be interviewed then they will sign a paper consent form to indicate what they consent to in terms of use of their data. These signed consent forms will be kept in a locked filing cabinet at WTSI.

All level 3 data (anonymous questionnaire data and video interview files) will be worked on from an encrypted, password protected computer as well as a portable device such as a laptop (also password protected and encrypted). There is a necessity to be able to work on the data on a portable machine because the researcher will be required to travel around the UK to collect the interviews, but also travel around the world presenting the project findings (including video clips) in the dissemination phase of the project. Between travelling it will be necessary to store the portable device at the researcher's home and this will be done in a locked,

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	<p>fire/bomb proof safe.</p> <p>The research participants are being informed, in detail, about their data being worked on from a portable device, and hence the risk that this may be stolen while the researcher is travelling. They consent specifically on the Consent Form to say they accept that there is a small risk that their interview (and thus their visual image) may become available if the laptop is stolen (and the password and encryption is successfully decoded). They will have previously had a chance to check what they said in their interview as we will send them a copy of this on DVD and so they can let us know if they are unhappy for any of this to be inadvertently become publically available.</p>
<p>14 What security checks do you have in place, e.g., encryption, locked filing cabinet, password protection?</p>	<p>Backup encryption of electronic files, password protected, locked filing cabinet.</p>
<p>15 How long will the project last?</p>	<p>2010-2015.</p> <p>Analysis of the data may continue for up to 15 years past the official end of the project. This analysis will involve the anonymous written transcripts from the interviews, the anonymous completed questionnaires and (if the research participant consented to this) the video files.</p>
<p>16 How long will the personal data be stored for?</p>	<p>The vast majority of personal data will have been destroyed before the end of the project (i.e. if a name and address was provided so that an interview could be completed then after the interview has been conducted the name and address will be destroyed).</p> <p>However, there may well be small numbers of personal data that is stored right up to the end of the project (i.e to 2015 when the project finishes). After this date all the provided personal data (names, addresses, email addresses) will be destroyed.</p> <p>If research participants consented to their data being used in future social sciences</p>

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	<p>research projects then a copy of their signed consent form (containing their name and signature, but no other identifiable information) will be stored and handed over to the next research team (who will need to obtain REC approval to work on this data).</p> <p>The video files obviously contain a visual image of the research participant, but there will be no name or other identifiable information attached to this. The video files will be stored, using the same Level 3 electronic protection as stipulated in the WTSI Data Storage guidance for as long as they are being analysed, including during the 15 years after the end of the project. The video files (with consent) will only be handed over to future researchers who can demonstrate that they have a similar level of data protection and storage policies to the WTSI.</p>
<p>17 How will all the types of identifiable data collected (e.g., electronic, video, paper) be destroyed?</p>	<p>The paper copies of the consent form will be shredded in 2015 unless the research participant gave specific consent to use their questionnaire and interview data in future social sciences research. In this case, the paper copies will be stored in a locked filing cabinet and then handed over to the next researchers.</p> <p>Electronic data (including avi files) will be destroyed as per the guidelines for Level 3 data.</p> <p>All level 4 data will be destroyed as per the guidelines for level 4 data.</p>
<p>18 What personal identifiers will you be using and why are these required?</p>	<p>Research participants can volunteer to provide their email address if they wish to receive details of the study results. They can also volunteer to have an interview, in which case they will choose to provide their name and address. We will be able to link a name and address to the responses given in the research questionnaire and to the interview; although once the interview is done, there is no need to store the name and address anywhere (indeed it will not appear on the written transcript created from the interview)</p>

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	<p>and thus can be destroyed.</p> <p>The questionnaire itself does contain some sensitive data as it includes questions relating to demographic data, including ethnicity.</p> <p>The questionnaire itself contains no embarrassing or sensitive data (it It asks for attitudes towards the sharing of genomic information for example and the research participant has filled it in willingly (i.e. they chose the answers that they gave), there is no predicted adverse outcome from there being a link between the research data and the identifiable information – this is quite a different scenario to molecular genetic data being linked to identifiable information.</p> <p>The personally identifiable information from the questionnaire will be separated out from the rest of the questionnaire responses and stored at Level 4. The rest of the questionnaire responses will be stored and managed at Level 3. The link between the personally identifiable data and the anonymous data will be stored at Level 4.</p>
<p>19 If storing personal identifiers alongside research data, please describe how personal identifiers, although linked to the data, will be anonymised. If the research data cannot be anonymised during the research process, please give reasons for this.</p>	<p>The online questionnaire data will go straight into an aggregate pool for analysis. It will not be possible, nor necessary, to identify individuals from this. Even though the original questionnaire that was completed may have contained a voluntarily provided email address at the end, this data will not be pooled into the aggregate file and will be stored separately as above. Thus all the questionnaire data will be completely anonymous.</p> <p>When the interviews are completed the audio/video file will be listened to and an anonymous written transcript will be created. Even if the research participant themselves provided some information in their interview that could be used to identify them, this will be deleted from the written transcript. The written transcript will be given a code and the transcript will be used in the analysis. Only the researcher (Anna Middleton) will have access to the key that links the identifiable information and the transcript. However,</p>

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	<p>once the interview has been completed there is no need to keep the names and addresses anymore and these will be deleted/destroyed.</p> <p>If the interview is audio-recorded the audio file will be destroyed after the written transcript has been created. If the interview is video-recorded and the research participant has not given consent for their video to be used in the dissemination phase of the project then once the written transcript has been created the video file will be destroyed. If they have given written consent for their video file to be used in the dissemination phase of the project or indeed in future research projects (that would require new REC approval) then these will be stored as above. No other identifiable information will be stored with the video files, such as name and address.</p>
<p>20 **Please describe the type of personal data you will be using, e.g., personal details, physical description, personality, family circumstances, social circumstances, education, physical health, mental health.</p>	<p>Email address Name Postal address</p> <p>Based on similar research of a similar size and scale done by the authors when based at Cardiff University (prior to working at WTSI) it is anticipated that there may be approximately 500 individuals who volunteer their email address, name and postal address.</p>
<p>21 If the results of the research will be shared with individuals or organisations directly associated with the Data Subject, please explain what data will be shared and with whom, e.g., the Data Subjects themselves, family, guardians, clinicians, legal representatives, social workers.</p>	<p>No individual research results will be shared with anyone directly associated with the data subject.</p>
<p>22 If the results of the research will be shared with individuals or organisations directly associated with the Data User, please explain what data will be shared and with whom, e.g., research sponsors, ethics committees, funding agencies, collaborators.</p>	<p>Aggregate (anonymous) results will be shared with research participants, research sponsors, funding agencies, research communities and collaborators.</p>
<p>23 If the results of the research will be shared with other individuals or organisations, please explain what data will be shared and with whom, e.g., Dept. of Health, health authorities, social services, central government, family</p>	<p>Aggregate (anonymous) questionnaire data will be shared publically with research participants, research sponsors, funding agencies, research communities and collaborators.</p>

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practitioner committees, hospitals, medical practitioners, health visitors, professional bodies.	Small sections of video may be shared with all of the above groups (if the research participant has given consent for this). Anonymous written quotes from individuals will be shared with all of the above groups.
24 Will the results of your research be published in an academic journal or other publication? Yes / No (Note: If you do publish the results of your research, they must not contain data by which an individual can be identified)	Yes All written publications will contain no identifiable information from research participants. All spoken conference presentations may contain video clips from the interviews (if the research participant consented to this).
25 Do you intend to transfer personal data to a country or territory outside the European Economic Area (EEA)? Yes / No (Note: If you do intend to transfer personal data outside the EEA, please give details and attach a copy of your agreement with the recipient)	No
Signature of Data User(s)	
Signature of Faculty Member	
For Office Use Only	
Signature of WTSI HMDMC Chair	ΕΝΕΥΘΕΡΙΑ ΖΕΡΓΙΝΗ
Date of Approval by Chair of WTSI HMDMC	12/02/2013

Notes

*The WTSI Human Genetics Data Security Policy can be found at: <https://helix.wtqc.org/services/human-genetics-data-security-policy>

**Further examples of personal data:

Personal characteristics	Personal details, physical description, habits, personality
Health and other classes	Physical health record, mental health record, disabilities and infirmities, dietary and other special health requirements, sexual life details, racial / ethnic origin, motoring and other convictions, criminal intelligence, political opinions, political party membership, support for pressure groups, religious or other beliefs
Family circumstances	Current marriage or partnership, marital history, details of other family or household members, other social contacts
Social circumstances	Accommodation or housing, immigration status, leisure activities, interests, membership of voluntary, charitable bodies, licences or permits held, court ; tribunal / inquiry proceedings, property, professions, travel details, lifestyle, public office held, complaint / incident / accident details
Education, Skills, profession	Academic record, qualification and skills, professional expertise, publications, student

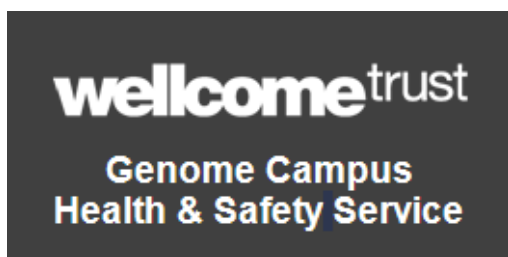
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Appendix C: Lone Worker Policy

Health & Safety Code of Practice

Home Visiting as Part of the DDD Project

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Controlled Document: **HSC**

Version:

Date:

Lead: sf10

Scope

Anyone involved in, or managing those undertaking home visits as part of the DDD project.

Definitions

Lone workers	Those who work by themselves without close or direct supervision.
Visitor	Member of WTSI staff who as part of their job role visit homes and premises of persons involved in research
Home visiting	Any visit to a third parties home or premises to carry out work relating to the DDD project. I.e. not the visiting persons' home address or usual place of work.

Objectives

To ensure that all persons engaged in home visits as part of the DDD project are not exposed to danger in accordance with the relevant legislation and guidance in this code of practice. This document assists in the identification of potential hazards and gives the basic safety precautions that must be adopted.

Arrangements

Members of the Board of Management are responsible for ensuring the implementation of this guidance in the areas under their control. Simply issuing this guidance to the concerned parties does not constitute implementation. Compliance should be achieved through the dissemination of information and the provision of appropriate training to all relevant persons.

They shall ensure that:

- That all users understand the hazards involved in home visiting;
- The necessary equipment and personal protective equipment (PPE) is provided;
- There are written emergency protocols;

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- The Campus Health and Safety Service is notified of any incidents;
- Sufficient resources are allocated to cover the above items.

Supervisors/Principal Investigators shall ensure that:

- A suitable and sufficient risk assessment is conducted; and a suitable procedure is produced.
- There is adequate training and, where necessary, supervision;
- Where training has taken place records are maintained for each individual.

Individual users shall ensure that:

- They take reasonable care of themselves and others affected by their actions;
- Local area procedures are complied with;
- That they do not endanger themselves or other by use of incorrect/ unsafe practices.
- They carry a **personal alarm** (to be kept in an accessible place).
- Every member of staff who travels away from the WTSI must complete a **visit proforma** (Appendix) as well as their personal diary as a means of logging visits. The visit proforma must be made easily available to colleagues who are monitoring your visit.
- Do not carry large amounts of **money or valuables**.
- New staff should have familiarisation sessions on home visits with an experienced member of staff during their induction period and only complete a home visit on their own when they, and their more experienced colleague, agree that they are ready. A formal risk assessment should be undertaken prior to new staff undertaking lone working.
- Obtain information about where you are visiting before the visit. Ask how many people will be at the visit.
- Risk assessment - ensure that there is opportunity to feedback relevant information from a lone visit - e.g. if you felt at risk or if there was an incident. This should be formally recorded and reviewed with your Manager/Supervisor and other members of your team to ensure appropriate follow up action is taken and to minimise any risk in subsequent visits.

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Pre - Visit

- Visiting staff must carry a formal means of identification.
- Ensure that the Visitor has detailed travel route instructions to their destination.
- Ensure that the Visitor has an effective means of communicating with their place of work or home.
- Make and keep pre-arranged appointments, and notify the participant if you cannot keep them.
- Arrange home visits during daylight hours whenever possible.
- When visiting the home of a participant, you should complete a visit proforma and leave the details with a nominated colleague or the Wellcome Trust Genome Campus Security Reception.
- Ensure that the nominated colleague or WTGC Security Reception are aware of the details of your visit and have agreed to monitor during the visit and when the visit is completed.
- Ensure that those persons nominated are available on the phone and contactable by you for the duration of your visit.
- Contact your nominated colleague if you are late for your appointment and ask them to note this on the visit proforma.
- Prepare yourself for difficult meetings by finding out everything you need to know before arriving and planning in your mind how you are going to deal with the situation.
- **Think carefully about the following procedures for ensuring your safety during home visits. Use your professional and personal judgement to decide whether they are appropriate to any given situation. If in doubt, adhere to the following guidelines:**

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During Visit

- **Do not** enter someone's home, if you don't feel comfortable or safe.
- **Do not** enter a house if the person you have arranged to see is not there. Be aware of, and maintain, personal safety at all times during visits.
- Always explain your research role clearly and the conditions of confidentiality.
- Your safety is the primary concern, which should be placed above completion of research tasks.
- Leave your mobile telephone switched on during the interview.

On Completion of a Visit

- The Visitor must contact their nominated colleague or the Wellcome Trust Genome Campus Security Reception as soon as they have completed the home visit. This time will be noted on the visit proforma.
- If the Visitor decides that they are not going to return to the office after their last visit, they should ensure that the appropriate person in the office knows about that by telephoning in. The visit pro-forma can then be completed accordingly.
- The researcher should contact a colleague at the office if an additional home visit is to be made and give all the relevant details which will be recorded on a new visit proforma. N.B. if this colleague is different from the nominated colleague from visit 1, then the researcher must ensure visit 1 nominated colleague is informed.
- It is the responsibility of the nominated colleague to ensure s/he is available to receive a call and monitor the time when the visit should be over. If circumstances change, s/he should arrange for another colleague to monitor the visit.
- If the interview is still in progress as the deadline for contacting the department approaches, the researcher should excuse him/herself and call their nominated colleague to inform them.

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- If the deadline passes and the researcher has not contacted the nominated colleague, the nominated colleague should ring the mobile telephone number of the researcher. If there is no answer, the nominated colleague should inform the Genome Campus Security immediately and ensure the police are informed immediately.

Home Visit Good Practice

- If you feel at all uneasy about conducting a home visit on your own, ask a colleague to accompany you.
- If you are late arriving for your appointment, let your nominated colleague/ Genome Campus Security Reception know and record the revised time on the visit proforma.
- When visiting people's home, try to let the tenant lead the way. Avoid being the first to go into any room. Be extra careful when alone with participants e.g. fetching something from a handbag, comforting participants. You should always make sure that the exit from the room is clear.
- Animals in the home: if you are in any doubt about the behaviour of animals in the home, ask for it/them to be locked away while you are visiting.
- Never undertake an interview or assessment in the bedroom.
- Do not give your personal telephone number or address.
- You should not interview anyone who is under the influence of alcohol or drugs.
- If you feel uncomfortable while in a person's home, you should take steps to leave immediately.
- A professional and friendly attitude should be adopted but over familiarity must be avoided.
- Remember that the interviewee may also feel anxious about the interview and your visit. You should bear this in mind whilst also ensuring your own safety.
- Be alert for signs of DANGER

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- Raised voice, rapid speech and babbling indicates rising tension.
- Changes in tone and pitch as the conversation progresses may suggest anger, frustration or impending violent behaviour.
- Keep your distance. Each of us has a personal space, which we defend when we feel it is being invaded.
- Be alert for body language that may indicate developing anger - e.g. flushed face, fidgeting, pointing, folded arms.
- Awkward or potentially threatening situations
- If an awkward or potentially threatening situation arises, this should be reported to a colleague as soon as possible. The facts should also be recorded in a specific "untoward incident" file.
- Formal arrangements should be in place for staff to be accompanied by a colleague for subsequent visits if there have been any incidents giving cause for concern on the first occasion.
- If, for any reason, you are concerned for your personal safety once you arrive at your appointment venue, then do feel able to cancel your appointment. On return to the office, make alternative arrangements when another member of staff experienced in working on their own undertaking home visits can accompany you.

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Home Visit Proforma

	Example Data
Name of person conducting visit off campus	Dr Anna Middleton
Mobile number of person conducting visit	0788 190 3069
Car Registration, make, model, colour	KT05 KFJ Mercedes, B-Class, grey
Name of person monitoring visit	Dr Caroline Wright (if before 5pm) or Alastair Gadney (husband) (if after 5pm)
Date of visit	Monday 6 th June 2011
Time of visit	2pm
Duration of visit	1 hour
Address of visit	3 Bulstrode Gdns Cambridge CB3 0EN
Gender of research participant	Male
Number of participants	1
Expected route	Google maps directions: CB10 1SA to CB3 0EN
Notes Person monitoring the visit should write additional information as necessary here (e.g. if the researcher calls to say they are late etc.)	

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Appendix D: Favourable REC Opinion



National Research Ethics Service NRES Committee East of England - Cambridge South

Victoria House
Capital Park
Fulbourn
Cambridge
CB21 5XB

Telephone: 01223 597656
Facsimile: 01223 597645

26 August 2011

Dr Anna Middleton
Ethics Researcher
Wellcome Trust Sanger Institute
Genome Campus
Hinxton
Cambridge
CB10 1SA

Dear Dr Middleton

Study title: Exploring the ethical implications of whole genome studies
REC reference: 11/EE/0313

Thank you for your letter of 23 August 2011 responding to the Proportionate Review Sub-Committee's request for changes to the documentation for the above study.

The revised documentation has been reviewed and approved by the sub-committee.

Confirmation of ethical opinion

On behalf of the Committee, I am pleased to confirm a favourable ethical opinion for the above research on the basis described in the application form, protocol and supporting documentation as revised.

Ethical review of research sites

The favourable opinion applies to all NHS sites taking part in the study, subject to management permission being obtained from the NHS/HSC R&D office prior to the start of the study (see "Conditions of the favourable opinion" below).

Conditions of the favourable opinion

The favourable opinion is subject to the following conditions being met prior to the start of the study.

Management permission or approval must be obtained from each host organisation prior to the start of the study at the site concerned.

Management permission ("R&D approval") should be sought from all NHS organisations involved in the study in accordance with NHS research governance arrangements.

Guidance on applying for NHS permission for research is available in the Integrated Research Application System or at <http://www.rdforum.nhs.uk>.

This Research Ethics Committee is an advisory committee to the East of England Strategic Health Authority.
The National Research Ethics Service (NRES) represents the NRES Directorate within the National Patient Safety Agency and Research Ethics Committees in England

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Where a NHS organisation's role in the study is limited to identifying and referring potential participants to research sites ("participant identification centre"), guidance should be sought from the R&D office on the information it requires to give permission for this activity.

For non-NHS sites, site management permission should be obtained in accordance with the procedures of the relevant host organisation.

Sponsors are not required to notify the Committee of approvals from host organisations.

It is the responsibility of the sponsor to ensure that all the conditions are complied with before the start of the study or its initiation at a particular site (as applicable).

You should notify the REC in writing once all conditions have been met (except for site approvals from host organisations) and provide copies of any revised documentation with updated version numbers. Confirmation should also be provided to host organisations together with relevant documentation.

Approved documents

The documents reviewed and approved by the Committee are:

<i>Document</i>	<i>Version</i>	<i>Date</i>
Advertisement	version 1	25 July 2011
Evidence of insurance or indemnity	insurance details	25 July 2011
Interview Schedules/Topic Guides	version 1	25 July 2011
Investigator CV	Dr Anna Middleton	
Other: WTSI Data Security Policy		25 July 2011
Other: Lone Working Guidance for Staff		25 July 2011
Other: Letter from NHS R & D - no approval required		25 July 2011
Participant Consent Form	version 2	23 August 2011
Participant Information Sheet: information booklet	version 2	23 August 2011
Protocol	version 1	21 July 2011
Questionnaire: Online Questionnaire	version 1	25 July 2011
REC application	78789/23494 2/1/275	
Response to Request for Further Information	from Anna Middleton	23 August 2011
Summary/Synopsis	Appendix A. Flow diagram of questionnaire design - version 1	25 July 2011

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

After ethical review

*This Research Ethics Committee is an advisory committee to East of England Strategic Health Authority
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Reporting requirements

The attached document "After ethical review – guidance for researchers" gives detailed guidance on reporting requirements for studies with a favourable opinion, including:

- Notifying substantial amendments
- Adding new sites and investigators
- Notification of serious breaches of the protocol
- Progress and safety reports
- Notifying the end of the study

The NRES website also provides guidance on these topics, which is updated in the light of changes in reporting requirements or procedures.

Feedback

You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.

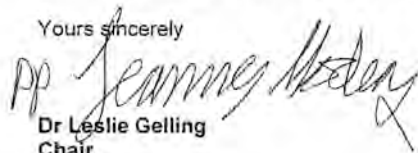
Further information is available at National Research Ethics Service website > After Review

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Please quote this number on all correspondence

With the Committee's best wishes for the success of this project

Yours sincerely



Dr Leslie Gelling
Chair

Email: leanne.moden@eoe.nhs.uk

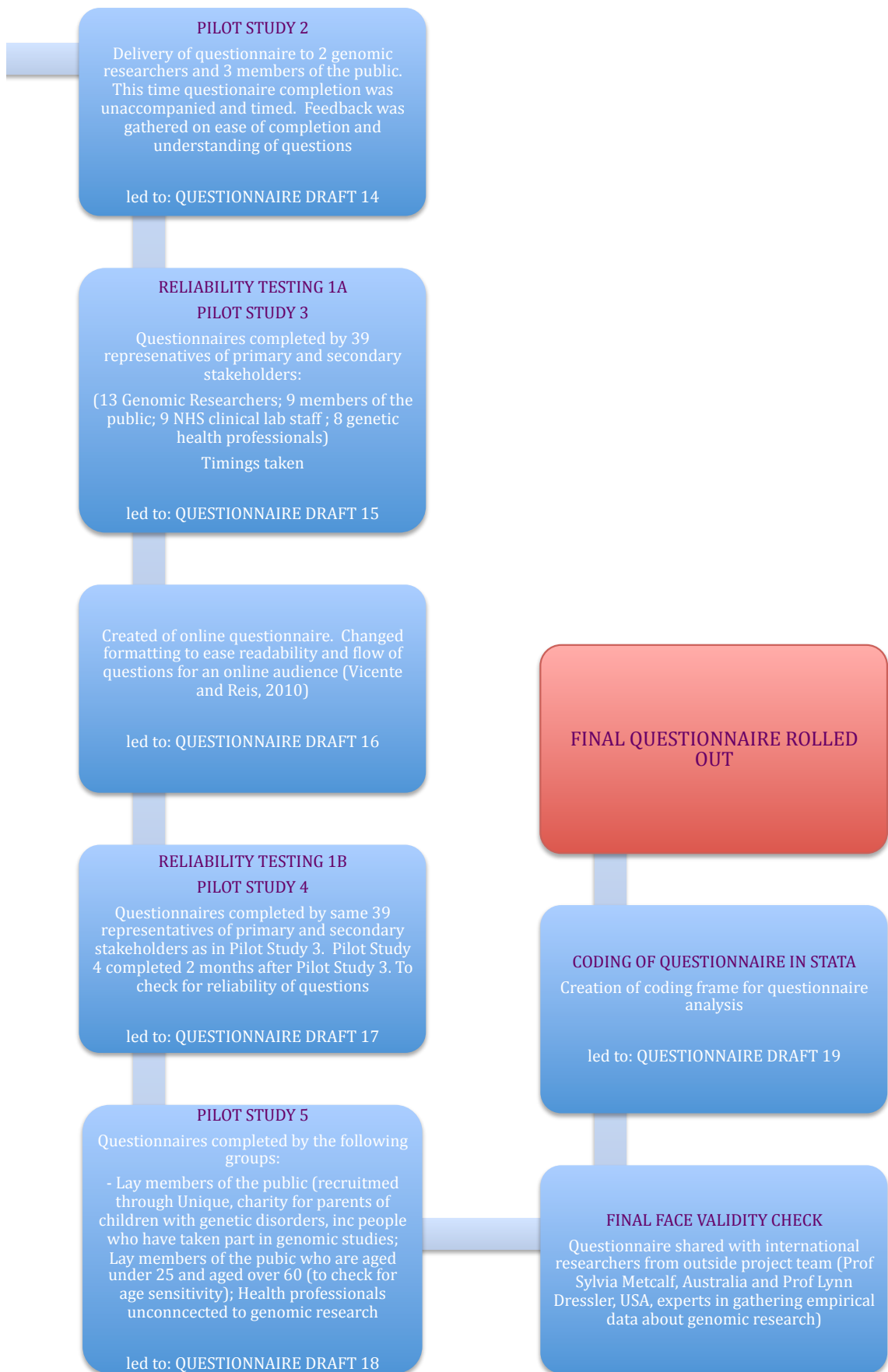
Enclosures: *After ethical review – guidance for researchers*

Copy to: *Dr David Davison*
Wellcome Trust Sanger Institute
Genome Campus
Hinxton
Cambridge
CB10 1SA

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the National Patient Safety Agency and Research Ethics Committees in England*

Appendix E: Creation of a Bespoke Survey

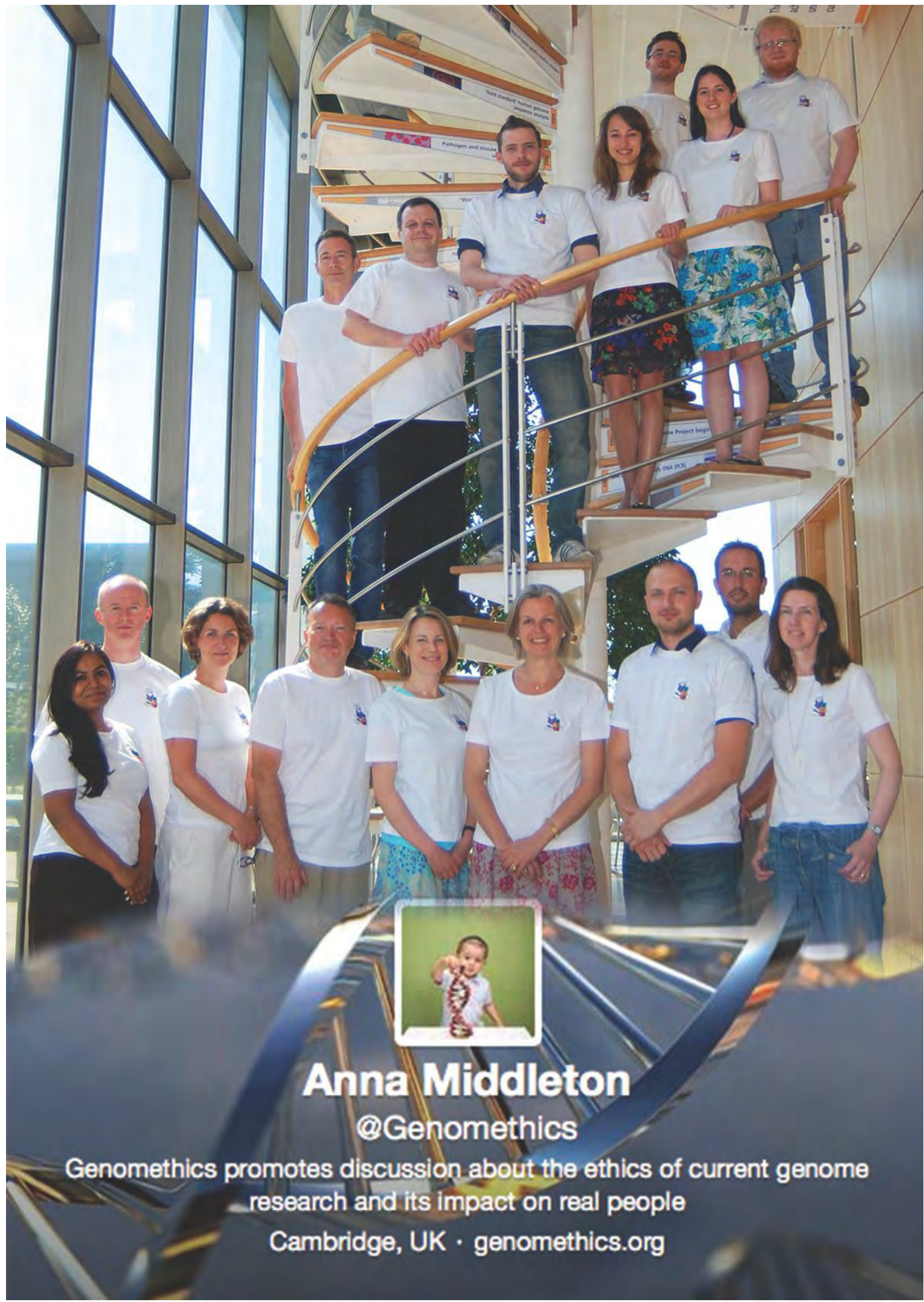




GENOMETHICS

DDD/Ethics Project





Anna Middleton

@Genomethics

Genomethics promotes discussion about the ethics of current genome research and its impact on real people

Cambridge, UK · genomethics.org