Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.

6th Plenary Programme Booklet

Congress Center Basel

Basel, Switzerland

October 3 – 5, 2018

ga4gh.org | @GA4GH | #GA4GH2018

Table of Contents

GA4GH 6th Plenary Objectives

The GA4GH 6th Plenary Meeting will present the first set of deliverables developed under GA4GH Connect — our 5-year strategic plan to enable real-world genomic data sharing by 2022. It will focus on real-world international use cases of GA4GH.

We are grateful for the support of our local meeting sponsors, the SIB Swiss Institute of Bioinformatics and ELIXIR, who will co-host the meeting.

Programme Committee

Kathryn North (Chair), Dixie Baker, Michael Baudis, David Glazer, Peter Goodhand, Robert Freimuth, Madeleine Murtagh, Gunnar Ratsch, Heidi Rehm, Serena Scollen, Torsten Schwede

Welcome to the GA4GH 6th Plenary Meeting

3 October, 2018

Dear Colleagues:

Welcome to Basel and to the 6th Plenary Meeting of the Global Alliance for Genomics and Health (GA4GH). We are delighted to have you here and anticipate a productive, inspiring three days. This year's programme is packed with great content and all of it is made possible thanks to the incredible support of our two local hosts: SIB – Swiss Institute of Bioinformatics and ELIXIR, the European life sciences infrastructure.

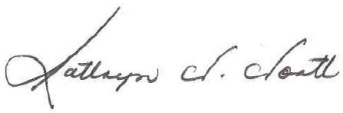
On day 1 a number of working meetings along with open technical and clinical workshops will let attendees dive right into the nitty gritty of genomic data sharing. GenoPri'18 will take place concurrently for those interested in genome privacy and security. On day 2 we will focus on the story of GA4GH so far, announcing the first four deliverables from the 2018 Strategic Roadmap. We will also discuss responsible sharing in an increasingly open world with invited talks and panels and watch a series of broadly-accessible demonstrations of a variety of tools either in development or ready for release. On our last day together, we will focus on "going global" and how national initiatives from around the world can help us make international genomic data sharing a reality.

Our keynote speakers — Andrew Morris (HDRUK), Torsten Schwede (University of Basel), and Nicola Mulder (University of Cape Town) — will address our two themes for the meeting: upholding a responsible framework for sharing data and sharing data across international bounds.

In this booklet, we hope you'll find answers to many of your questions, including details about the programme, venue, and local region. If you need anything else during your time in Basel, please do not hesitate to reach out. The registration booth will be staffed throughout the meeting, and you can always reach us on Twitter (@GA4GH) or via email (info@ga4gh.org).

Finally, I would like to thank all of the GA4GH 2018 Funding Partners, Core Funders, and Host Institutions who together make this meeting possible. Without the continued support of these organizations and agencies, the important work GA4GH is doing to advance genomic data sharing could not happen. Please see a full list of funders and supporters on p.5.

Warm regards,



Kathryn North, Chair of the Scientific Programme Committee
Member, GA4GH Executive Committee
Director, Murdoch Childrens Research Institute
Director, Australian Genomics

Message from the GA4GH Steering Committee Chair

3 October, 2018

Dear Colleagues:

I am thrilled to be here in Basel with you as we learn together about the impressive work of the GA4GH Work Streams. They are developing a suite of practical deliverables to overcome the technical challenges of sharing genomic and health related data that will help all of us as we work to advance precision medicine. We also have organised an open and inclusive agenda of other leaders and groups in the genomics and biological standards arena to help bridge GA4GH to related work around the world.

In the latter part of 2017, directly following 5th Plenary, members of the Work Streams met with each of our 15 inaugural Driver Projects to come up with a strategic roadmap consisting of 28 unique deliverables that tackle everything from the discovery of data, to access and transfer, and through to mechanisms for secure cloud-based storage and analysis.

This roadmap is incredibly ambitious and our active contributors have been labouring tirelessly over the past 12 months to bring it to life. The first set of deliverables to come of this effort will be released on October 4 during the session entitled, “GA4GH Connect: The Story So Far”. We will learn all about the new deliverables, as well as several others expected to be released in the coming months, during talks and demos.

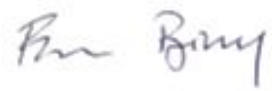
You can learn more about the details of these APIs on the GA4GH GitHub repository github.com/ga4gh as well as on our website, ga4gh.org. In addition, high level summaries can be found in the pages of this booklet.

I cannot tell you how pleased I am at the great work of this group. I hope you’ll join me in congratulating them on these first successes as we look toward the next phase of this journey, which will be to ensure their application around the globe to advance data sharing.

Toward that end, I also want to draw your attention to the events on October 5, when we will issue an open call for new Driver Projects with an eye toward increased global representation.

Thank for joining us in Basel. I look forward to spending the next three days with you.

Best wishes,



Ewan Birney, GA4GH Chair
Director, European Bioinformatics Institute (EMBL-EBI)

Funders and Sponsors

A big thank you to our GA4GH 6th Plenary Co-Hosts, and Funding Partners for their generous support!

Local Partners and Co-Hosts

Funding Partners

GA4GH 6th Plenary: Day 1

Wednesday, October 3, 2018
Basel, Switzerland

GenoPri
8:10 - 17:00 | *Singapore Hall*
Agenda at www.genopri.org

Registration Opens
7:00am

11:00

WELCOME & OPENING REMARKS
Montreal Room

Ewan Birney (EMBL-EBI, GA4GH Chair)

11:30

INTRO TO GA4GH CONNECT
Montreal Room

Peter Goodhand (OICR, GA4GH), **Adrian Thorogood** (GA4GH), **Melissa Konopko** (GA4GH), **Rishi Nag** (GA4GH)

13:00

LUNCH
2nd Floor Foyer

14:00

WORKSHOP: GENOMIC DATA COMPRESSION
Sydney Hall

Chairs:
Thomas Keane (EMBL-EBI),
Marco Mattavelli (École polytechnique fédérale de Lausanne)

GENE WORKSHOP: TRANSCRIPT CHOICE
Osaka Room

Chairs:
Andy Yates (EMBL-EBI),
Robert Freimuth (Mayo Clinic),
Fiona Cunningham (EMBL-EBI)

ELIXIR Meeting
Samarkand Room

Chairs:
Serena Scollen (ELIXIR),
Gary Saunders (ELIXIR),
Michael Baudis (University of Zurich)

15:30

BREAK

16:00

WORKSHOP: GENOMIC DATA COMPRESSION (continued)
Sydney Hall

Chairs:
Thomas Keane (EMBL-EBI),
Marco Mattavelli (École polytechnique fédérale de Lausanne)

WORKSHOP: DEMONSTRATING THE IMPACT OF GENOMIC DATA SHARING
Montreal Room

Chairs:
Kathryn North (Murdoch Childrens Research Institute),
Anna Middleton (Wellcome Sanger Institute)

GENE WORKSHOP: GENE-DISEASE VALIDITY
Osaka Room

Chair:
Heidi Rehm (Broad Institute),
Ellen McDonagh (Genomics England)

ELIXIR Beacons Workshop
Samarkand Room

Serena Scollen (ELIXIR),
Gary Saunders (ELIXIR),
Michael Baudis (University of Zurich)

17:30

NETWORKING COCKTAIL
2nd Floor Foyer

18:30

GA4GH 6th Plenary: Day 2

Thursday, October 4, 2018
Basel, Switzerland

Registration Opens
7:00am

8:00 **WORKSHOP: NATIONAL INITIATIVES DISCUSSION**
Singapore Hall
Chairs
Kathryn North (Murdoch Childrens Research Institute), **Mark Caulfield** (Genomics England)

9:30 **BREAK**

10:00 **OPENING REMARKS: Ewan Birney** (EMBL-EBI, GA4GH Chair)
Montreal Room

10:15 **GA4GH CONNECT: THE STORY SO FAR**
Montreal Room
Chair: Ewan Birney (EMBL-EBI, GA4GH Chair)
Speakers: Mike Lin (DNAnexus), **Thomas Keane** (EMBL-EBI), Andy Yates (EMBL-EBI), **David Glazer** (Verily), **Harindra Arachchi** (Broad Institute), **Alex Wagner** (Washington University), **Dixie Baker** (Martin, Blanck & Associates), **Kate Birch** (Australian Genomics), **Madeleine Murtagh** (University of Edinburgh), **Moran Cabili** (Broad Institute), **Melanie Courtot** (EMBL-EBI), **Julius Jacobsen** (Queen Mary University of London), **Heidi Rehm** (Broad Institute), **Marc Fiume** (DNAstack), **Stephanie Dyke** (McGill), **Ilia Tulchinsky** (Google Cloud)

13:00 **LUNCH**
2nd Floor Foyer
Attend an open town hall meeting organized by the National Human Genome Research Institute to discuss genomics research beyond 2020.
A separate lunch station will be set up for those who wish to join. Sign up here.

14:00 **KEYNOTE ADDRESS: RESPONSIBLE DATA SHARING**
Montreal Room
Andrew Morris (Health Data Research UK)
Introduction: Mark Caulfield (Genomics England)

14:45 **RESPONSIBLE GENOMIC DATA SHARING PANEL: RIGHT TO BENEFIT**
Montreal Room
Chair: Madeleine Murtagh (University of Edinburgh)
Speakers: Anna Middleton (Wellcome Sanger Institute), **Peter Lichter** (German Cancer Research Center), **Rumiana Yotova** (University of Cambridge)

15:45 **BREAK**

16:15 **RESPONSIBLE GENOMIC DATA SHARING PANEL: PRIVACY & SECURITY**
Montreal Room
Chair: Paul Flicek (EMBL-EBI)
Speakers: Jean-Pierre Hubaux (École Polytechnique Fédérale de Lausanne), **Alessandro Blasimme** (ETH Zurich), **Michaela Mayrhofer** (BBMRI)

17:20 **NETWORKING DINNER**
2nd Floor Foyer

19:05

GA4GH 6th Plenary: Day 3

Friday, October 5, 2018
Basel, Switzerland

Registration Opens
7:30am

8:30	**OPENING REMARKS** *Montreal Room* **Ewan Birney** (EMBL-EBI), **Peter Goodhand** (GA4GH)
8:45	**KEYNOTE ADDRESS: GOING GLOBAL** *Montreal Room* **Torsten Schwede** (Swiss Personalized Health Network) **Nicola Mulder** (H3Africa, University of Cape Town) **Introductions: Ewan Birney** (EMBL-EBI)
9:55	**PANEL: PATIENT PERSPECTIVES** *Montreal Room* **Chair: Heidi Rehm** (Broad Institute) **Speaker: Peter Kapitein** (Inspire2Live), **Olivier Menzel** (BLACKSWAN Foundation), **Corrie Painter** (Broad Institute)
11:00	**BREAK**
11:30	**PANEL: ADVANCING STANDARDS IN GENOMIC AND HEALTH DATA** *Montreal Room* **Chair: Ewan Birney** (EMBL-EBI) **Speakers: Robert Freimuth** (HL7), **Ian Green** (SNOMED), **Peter Van Reusel** (CDISC), **Bron Kisler** (ISO), **Christine Bakan** (Roche), **Augusto Rendon** (Genomics England)
12:45	**LUNCH** *2nd Floor Foyer*
13:45	**PANEL: NATIONAL INITIATIVES** *Montreal Room* **Chair: Kathryn North** (Murdoch Childrens Research Institute) **Speakers: Kathryn North** (Murdoch Childrens Research Institute), **Mark Caulfield** (Genomics England), **Chris Lunt** (All of Us), **Hidewaki Nakagawa** (Japan Agency for Medical Research and Development), **Anne Cambon-Thomsen** (Plan France Médecine Génomique 2025), **Ilkka Lappalainen** (ELIXIR)
15:30	**CLOSING REMARKS** *Montreal Room* **Ewan Birney** (EMBL-EBI)
15:45	**Meeting Adjourns**
16:00	**EGA 10 YEAR SYMPOSIUM** *Singapore Hall* Hosted by EMBL-EBI Registration is now closed for the Symposium. Please find the agenda here.

5th International Workshop on Genome Privacy and Security (GenoPri'18)

OCTOBER 3, 2018, 8:10 - 17:00

SINGAPORE HALL
CONGRESS CENTER BASEL

KEYNOTE SPEAKERS

Human genomic data sharing – A perspective for the next decade
Ewan Birney
European Bioinformatics Institute

Security in Personal Genomics: Lest We Forget
Gene Tsudik
University of California, Irvine

View the full program at genopri.org/program

#Genopri18

Subscribe to Our Monthly GDPR Briefs!

GA4GH GDPR and International Health Data Sharing Forum

Forum Editors

Edward Dove (University of Edinburgh)

Mark Phillips (McGill University)

A new forum supported by the GA4GH Regulatory and Ethics Work Stream will publish monthly briefs that answer important questions about the impact of the European Union's General Data Protection Regulation on various aspects of international health research and genomic and health-related data sharing.

- ➔ Subscribe to our monthly briefs: https://goo.gl/CbQP6x
- ➔ Read the introductory Primer: http://goo.gl/aPQ8iM

Overview of Day 1 Workshops

GenoPri '18
08:00 - 17:00
Singapore Hall

CHAIRS: Dixie Baker (Martin Blanck & Associates), Emiliano De Cristofaro (University College London)

Join the international data privacy community for a day of talks on public perceptions of sharing, privacy protections, governance and more.

Learn More

Workshop: Demonstrating the Impact of Genomic Data Sharing
16:00 - 17:30
Montreal Room

CHAIRS: Kathryn North (Australian Genomics), Anna Middleton (Wellcome Sanger Institute)

An open discussion to share and promote "good news stories" of genomic data sharing.

Share a story

Gene Workshop: Transcript Choice
14:00 - 15:30
Osaka Room

CHAIRS: Andy Yates (EMBL-EBI), Robert Freimuth (Mayo Clinic), Fiona Cunningham (EMBL-EBI)

Discussion of transcript harmonisation work that the EMBL-EBI and NCBI are proposing and current progress.

Learn More

Workshop on Genomic Data Compression
14:00 - 17:30
Sydney Hall

CHAIRS: Thomas Keane (EMBL-EBI), Marco Mattavelli (École polytechnique fédérale de Lausanne)

Come learn about and discuss the suitability of new and emerging compression technologies to address the specific challenges of large scale genomic data.

Learn More

ELIXIR Meeting / ELIXIR Beacons Workshop
14:00 - 17:30
Samarkand Room

CHAIRS: Serena Scollen (ELIXIR), Gary Saunders (ELIXIR), Michael Baudis (University of Zurich)

An open meeting for ELIXIR representatives and others to learn about areas of alignment between GA4GH and the European life sciences network, ELIXIR.

Learn More

Gene Curation Workshop: Gene-Disease Validation
16:00 - 17:30
Osaka Room

CHAIRS: Heidi Rehm (Broad Institute), Ellen McDonagh (Genomics England)

To align all groups defining the validity of gene-based relationships, which inform their use in clinical testing, including the formation of gene panels.

Learn More

Calling All National Initiatives!

Please bring your breakfast and join us for a breakout session prior to opening remarks on Day 2 of the GA4GH 6th Plenary Meeting to discuss key challenges across national and international genomic data initiatives, share resources, and learn from one another.

National Initiatives Workshop

October 4

08:00-09:30

Singapore Hall, Congress Centre Basel

To make the most of the precision medicine promise, we must work together to compare experiences and learn from one another — not reinvent the wheel each time one of us takes on a new data sharing challenge.

Hosted By:

Kathryn North (Australian Genomics)

Mark Caulfield (Genomics England)

NHGRI Town Hall Meeting

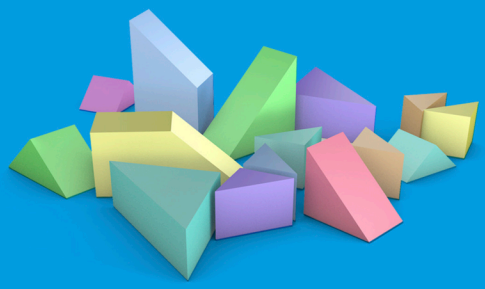
October 4, 2018
13:00 - 14:00
Singapore Hall

Please join the National Human Genome Research Institute (NHGRI) (NIH, USA) for an open town hall lunch meeting to discuss your ideas for the most exciting opportunities for genomic research and its applications in health and disease.

A separate lunch station will be set up for those who wish to join.

Please Register Here:
goo.gl/2tQBWX

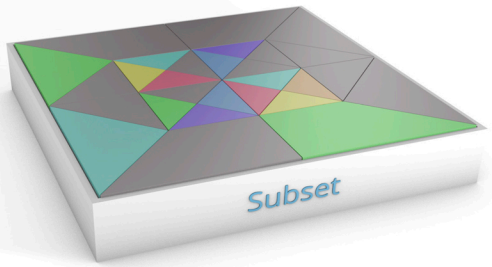
The GA4GH Suite:
Standards for Genomic Data Sharing



discovery | access | transfer | storage | analysis | cloud

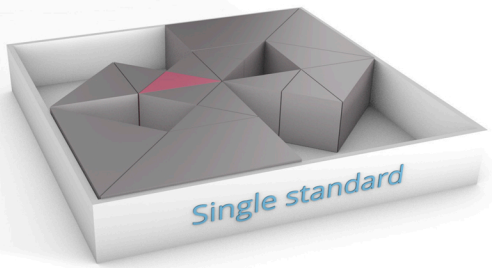
Four approaches to implementing GA4GH standards:

Use the **full suite** for an end-to-end solution to genomic data sharing



Use a **subset** to tackle a handful of specific data sharing challenges

Use **one standard** to fill a gap in an existing workflow



Use **one standard** alongside other solutions as you develop a new workflow

Visit the GA4GH website to find out more: www.GA4GH.org

A new standard for case discovery...

The GA4GH Search API

Towards a global federated data sharing network that allows you to query and — if you want to — process the results in the cloud.

Building on the Matchmaker Exchange API, the search API allows you to search without a pre-existing patient record — you can query anything you wish.

Implementations:

- CaseLog (Illumina)
- VariantMatcher (Johns Hopkins University)
- Cafe Variome (University of Leicester)

Access reference genomic sequences from different databases and servers using a checksum identifier unique to the sequence itself.

The GA4GH refget API

DID YOU KNOW?

refget forms a fundamental component of the CRAM file format.

FIND OUT MORE

For more information please visit goo.gl/QaMRC4

Lighting the Future for Variant Data Discovery

Light a Beacon

Present a genomic data collection as a web service, allowing users to query for the presence or absence of a particular allele without disclosing any further data differentiating the individuals.

Let Beacon Light Your Way:

Beacon Network

Find datasets around the globe that contain a variant of interest to you (consists of a subset of all “Beaconized” datasets)

ELIXIR Beacon Network

Find datasets in the European life sciences infrastructure, ELIXIR, that contain a variant of interest to you

Individual Beacons

Determine whether a particular dataset contains an allele of interest

Calling All Molecular Geneticists!

If you are interested in helping us spread the word about this important resource among your colleagues, please get in touch: info@ga4gh.org

WES

The Workflow Execution Service API

Run a single workflow on multiple platforms, clouds, and environments using either Common Workflow Language (CWL) or Workflow Definition Language (WDL).

Key features of the API

- Request a workflow to be run
- Pass parameters to that workflow
- Get information about running workflows
- Cancel a running workflow

More info:

ga4gh.github.io/workflow-execution-service-schemas/docs/

HTSGET *(V.1.0.0)*

GITHUB REPOSITORY

https://github.com/samtools/hts-specs

DOCUMENTATION

https://samtools.github.io/hts-specs/htsget.html

htsget (v.1.0.0) is a genomic data retrieval specification that allows users to download read data for subsections of the genome in which they are interested. Currently, users must download the whole set of files in which those data reside, a slow, resource-intense process.

CONTACT

To learn more, contact
GA4GH Technical Programme Manager
Rishi Nag @ rishi.nag@ga4gh.org

You know the file formats....

- SAM/BAM (for storing read data)
- CRAM (for more efficient storage of read data with better lossless compression)
- VCF/BCF (for storing variant data)

But do you know the File Formats Task Team?

This subgroup of the GA4GH Large Scale Genomics Work Stream is the international maintainer of the genomic data file formats you use in your work all the time. Contact the team members below to learn more.

Contact Us to Learn more:

Rishi Nag
Technical Programme Manager

James Bonfield
File Formats Lead

Thomas Keane
File Formats Lead

Keynote Speakers

NICOLA MULDER

University of Cape Town, South Africa

Nicola Mulder heads the Computational Biology Division at the University of Cape Town, and leads H3ABioNet, a large Pan African Bioinformatics Network of 27 institutions in 16 African countries. H3ABioNet aims to develop bioinformatics capacity to enable genomic data analysis on the continent by developing and providing access to and skills and computing infrastructure for data analysis. Prior to her position at UCT, she worked for 9 years at the European Bioinformatics Institute (EBI) in Cambridge, as a Team Leader, responsible for the development of InterPro, a heavily used bioinformatics resource at the Institute. At UCT her research focuses on genetic determinants of susceptibility to disease, African genome variation, and microbial genomics and infectious diseases from both the host and pathogen perspectives. Her group also provides bioinformatics services for the local researchers, through which they develop visualization and analysis tools for high-throughput biology. Her team led the design of the H3Africa genotyping array and has also been involved in the development of new and improved algorithms for the analysis of complex African genetic data as well as for downstream analysis and interpretation of GWAS data. Prof Mulder is actively involved in training and education as well as curriculum development in Bioinformatics and Genomic Medicine.

ANDREW MORRIS

Health Data Research UK, United Kingdom

Andrew Morris is the inaugural Director of Health Data Research UK, the multi-funder UK Institute for health and biomedical informatics research that will capitalise on the UK's renowned data resources and research strengths to transform lives through health data science. He is seconded from his position as Professor of Medicine, and Vice Principal of Data Science at the University of Edinburgh, having taken up position in August 2014. Prior to this Andrew was Dean of Medicine at the University of Dundee.

Andrew was Chief Scientist at the Scottish Government Health Directorate (2012-2017) and has served and chaired numerous national and international grant committees and Governmental bodies.

Andrew was awarded a CBE (Commander of the Most Excellent Order of the British Empire) in the 2018 New Year's Honour's List.

His research interests span informatics and chronic diseases. He has published over 300 original papers and has attracted over £50million in grant funding.

TORSTEN SCHWEDE

University of Basel, Switzerland

Torsten Schwede studied Chemistry in Bayreuth and completed his PhD in Structural Biology in Freiburg, Germany. He then worked as a Bioinformatics Scientist at GlaxoWellcome in Geneva (later GSK) before he was appointed tenure-track assistant professor of Bioinformatics at the Biozentrum in Basel in 2001.

Since 2002, Torsten has been a group leader at the SIB Swiss Institute of Bioinformatics. In 2007, he was promoted to Associate Professor and, in 2014 he became a Director of sciCORE, where he is responsible for the central infrastructure for scientific computing at the University of Basel. In 2018 Torsten has been appointed the Vice President for Research at the University of Basel. As a chairman of the Scientific Expert Board and a director of the Data Coordination Centre of the Swiss Personalized Health Network (SPHN), Torsten is committed to the development of research in the field of personalized health and medicine in Switzerland.

Plenary Speakers

Harindra Arachchi
Broad Institute, USA

Harindra Arachchi, MBA, is Co-Lead of the Discovery Work Stream (DWS) and a member of the steering committee of the Global Alliance for Genomics and Health (GA4GH). During his career at the Broad Institute, he has contributed technology, leadership, and scientific insight, to many large-scale genomics initiatives starting from the finishing of the Human Genome Project, to various others such as the Human Microbiome Project, and The Cancer Genome Atlas (TCGA). In addition to Broad, he has worked at the Whitehead Institute as well as the Pasteur Institute in Paris, France, where he helped launch a centralized big data consulting and engineering organization to help scientists work with large-scale next generation genomics data. Harindra has been part of many open source initiatives such as a leader in the Matchmaker Exchange (MME) technical working group, as well as leading the GA4GH Search-API, an important product in the strategic roadmap. The Search-API will help solve difficult rare disease, and other cases, by joining together disparate genomic data collections to empower research and clinical applications.

Christine Bakan
Roche Sequencing Solutions, USA

Christine Bakan is Global Head and Vice President of Software and Bioinformatics for Roche Sequencing Solutions. Christine leads research and development of genomic sequencing data analysis and clinical interpretation software for end-to-end, next generation sequencing solutions being developed at Roche.

Prior to Roche Sequencing Solutions, Christine held senior positions at Cisco Systems where she was responsible for core software technologies, digital network solutions and software defined networking across Cisco's market leading enterprise switching, routing and mobile/ wireless product lines. Christine previously led software research and development organizations for cloud IT service management products at BMC Software (top rated in Gartner Magic Quadrant), and software development teams in the advanced technology group at Oracle Corporation. She holds four patents related to virtualization, security and performance optimization in Oracle applications and database technologies.

Christine holds degrees in Molecular Biology and Neuroscience from UC Berkeley and in business from the Stanford University Graduate School of Business.

Dixie Baker
Martin, Blanck & Associates, USA

Dixie Baker is a Senior Partner at healthcare consulting firm Martin, Blanck and Associates, where she provides consulting services in the areas of health information technology (HIT), electronic health records (EHR) technology, privacy and security technology, and the sharing and protection of genomic and clinical data. Dr. Baker co-chairs the Security Foundational Work Stream of the Global Alliance for Genomics and Health (GA4GH), a worldwide coalition to facilitate the sharing of genomic and clinical data to accelerate the discovery and translation of biomedical knowledge. She serves on the Advisory Council for Health Level Seven International (HL7), the ANSI-accredited healthcare standards development organization, and on the Scientific Advisory Board of the European Genome-Phenome Archive (EGA). She also serves on the Institutional Review Board for Genetic Alliance. and on the U.S. Institute of Medicine's DIGITizE Action Collaborative, which is developing standards for integrating genomic data into Electronic Health Records. In 2009, Dr. Baker was named to the newly-formed U.S. Department of Health and Human Services HIT Standards Committee (HITSC) charged with recommending standards for EHR technology certification. She chaired the HITSC Security Workgroup throughout her 6+ year tenure, and led the Nationwide Health Information Network (NwHIN) Power Team that developed metrics for assessing the readiness of technology specifications to become national standards. She is a Fellow of the Healthcare Information and Management Systems Society (HIMSS). In 2017, Health Data Management named Dr. Baker one of the "most powerful women in health information technology," specifically in the area of "thought leadership." In 2013, healthcareinfosecurity.com selected her as one of its inaugural "Top 10 Influencers in Health Information Security."

Kate Birch
Melbourne Genomics Health Alliance, Australia

Kate Birch leads the Melbourne Genomics Health Alliance Data & Technology Program.

Kate's project delivery and research career has taken a 'bench to bedside' trajectory. She began as a molecular biologist, moving into epidemiological and then clinical research, and then clinical project delivery. She has consulted to the health industry with Deloitte, has executive level experience in a clinical research centre, and has an extensive working knowledge of medical research and hospital-based information management requirements, barriers and opportunities. Kate has worked in complex stakeholder environments to develop information system strategy, design and implement data governance activities, and to curate, manage and integrate large medical research datasets.

Kate has a Bachelor of Science (Honours), a Masters in Information Systems and is a Certified Health Informatician (Australasia).

Ewan Birney
EMBL-EBI, United Kingdom

Ewan Birney is Director of EMBL-EBI with Dr Rolf Apweiler, and runs a small research group. He is also EMBL-EBI's Joint Head of Research, alongside Dr Nick Goldman.

Ewan completed his PhD at the Wellcome Sanger Institute with Richard Durbin. In 2000, he became Head of Nucleotide data at EMBL-EBI and in 2012 he took on the role of Associate Director at the institute. He became Director of EMBL-EBI in 2015. Ewan led the analysis of the Human Genome gene set, mouse and chicken genomes and the ENCODE project, focusing on non-coding elements of the human genome. Ewan's main areas of research include functional genomics, DNA algorithms, statistical methods to analyse genomic information (in particular information associated with individual differences in humans and Medaka fish) and use of images for chromatin structure.

Ewan is a non-executive Director of Genomics England, and a consultant and advisor to a number of companies, including Oxford Nanopore Technologies, Dovetail Genomics and GSK. Ewan was elected an EMBO member in 2012, a Fellow of the Royal Society in 2014 and a Fellow of the Academy of Medical Sciences in 2015.

Alessandro Blasimme
Swiss Federal Institute of Technology (ETH), Switzerland

Alessandro Blasimme is a senior researcher in the Health Ethics and Policy Lab at the Department of Health Sciences and Technology of ETH Zurich, the Swiss Federal Polytechnic. His research focuses on innovation in biomedicine and on its ethical, regulatory and political consequences. In particular, his work has explored ethical issues in domains such as genomics, regenerative medicine, genome editing, precision medicine, digital health and aging research.

Prior to moving to ETH, Dr. Blasimme held research appointments at the French National Institute of Health and Medical Research (INSERM), and the University of Zurich, and was a Fulbright visiting scholar at Harvard University.

He received a degree in philosophy and a master in bioethics from "La Sapienza" University of Rome (Italy), as well as a PhD in Bioethics form the University of Milan (Italy) - European School of Molecular Medicine (SEMM).

Moran Cabili
Broad Institute, USA

Moran Cabili is a computational biologist and bioinformatician by training, passionate about solving problems to accelerate the impact of genomics on medicine. In her current role as a senior product manager at the Broad Institute Moran leads the development of software products that introduce new paradigms for sharing human genomic data to promote clinical and biomedical research. Moran also co-chairs the Data Use work-stream at the Global Alliance for Genomics and Health (GA4GH), focusing on standardization and automation of the human subject's data use oversight process. Moran received her PhD in Systems Biology from Harvard as an HHMI International Student Research Fellow and holds a MSC and BSC in computer science and life sciences from Tel-Aviv University.

Anne Cambon-Thomsen
National Center for Scientific research (CNRS), France

Anne Cambon-Thomsen, MD, immunogeneticist, specialised in human biology and health ethics, is Emeritus Research Director at the National Center for Scientific Research (CNRS) in a joint Unit on epidemiology and public health of the National Institute of health and medical research (Inserm) and University of Toulouse, France. She is member of the European Group on Ethics of Science and New Technologies (EGE) and co-pilots the working group « Ethics, regulation, society » of the French plan « Genomic medicine 2025 ». Previously she worked on human genetic variation in populations and diseases, transplantation immunogenetics, then societal and policy aspects of genomics and biotechnologies. She entered Inserm in 1976, completed a post-doctorate in Denmark, became research director at CNRS in 88. She directed two immunology and genetics research units (85-97), then led an interdisciplinary team on societal aspects of biotechnology for health and a platform of societal dialogue (1998-2016) in Toulouse. With experience in national and European ethics bodies and projects on ethical, legal and social implications (ELSI) of research, she co-directed the Common Service ELSI of BBMRI-ERIC, the European infrastructure on biobanking and biomolecular resources 2015-17 and is ESOF 2018 Champion: EuroScience Open Forum, the largest interdisciplinary science event in Europe.

Mark Caulfield
Queens Mary, University of London, United Kingdom

Mark Caulfield graduated in Medicine in 1984 from the London Hospital Medical College and trained in Clinical Pharmacology at St Bartholomew's Hospital where he developed a research programme in molecular genetics of hypertension which has discovered over 1000 gene loci for blood pressure and clinical research. He served on the NICE Guideline Group for hypertension and was President of the British Hypertension Society (2009-2011).

He was appointed Director of the William Harvey Research Institute in 2002 and was elected a Fellow of the Academy of Medical Sciences in 2008. He led on fundraising towards the £25m William Harvey Heart Centre which created a translational clinical research centre. Since 2008 he directs the National Institute for Health Research Cardiovascular Biomedical Research Unit and now Centre at Barts. Between 2010 and 2015 he co-led the merger of three hospitals in North London to create the new £400 million Barts Heart Centre which provides 80,000 cardiovascular patient episodes.

He has won the Lily Prize of the British Pharmacology Society, the Bjorn Folkow Award of the European Society of Hypertension 2016 and the Franz Volhard Award of the International Society of Hypertension in 2018.

In 2013 he became an NIHR Senior Investigator and was appointed as the Chief Scientist for Genomics England (100,000 Genomes Project) 2013-ongoing.

Melanie Courtot
EMBL-EBI, United Kingdom

Melanie Courtot leads the BioSamples project at EMBL-EBI. She is interested in making data available in a structured and standardised way, to help answer complex integrative queries and computationally process information. Melanie develops several ontologies and is an operations committee member of the OBO Foundry, a consortium of ontology developers committed to collaboration and adherence to shared principles. In her talk, Melanie will describe how data sharing policies can be formally expressed using the data Use Ontology, thus enabling automated matching between controlled-access datasets and data access requests. She will showcase an implementation between the European Phenome Genome archive and the Wellcome Trust Sanger Institute Data Access Committee.

Fiona Cunningham
EMBL-EBI, United Kingdom

Fiona Cunningham is interested in problems where building large-scale systems for genomic data can lead to fundamental biological insights, particularly for understanding genomic variation. Her team developed Ensembl's *Variant Effect predictor (VEP)*, the software tool for annotating variation data.

Fiona is part of the senior management for Ensembl (www.ensembl.org), a genome information system, and led the creation of the variation and phenotype data in Ensembl. In collaboration with the NHGRI, she is one of the PIs for the GWAS Catalog and she co-leads the Variant Annotation task team for the Global Alliance for Genomic Health project (GA4GH).

Fiona is committed to open data and standards, in particular to share data and facilitate the transfer of research knowledge into valuable resources. In collaboration with the NCBI in the USA, Fiona leads the development of reference sequences, called LRG (Locus Reference Genomic) sequences, a stable data standard for reporting clinical variants with support for legacy sequences.

Prior to EMBL-EBI, Dr Cunningham worked at the Wellcome Trust Sanger Institute, Cold Spring Harbor Laboratory, and at deCODE Genetics in Iceland.

Stephanie Dyke
McGill University, Canada

Stephanie Dyke's research experience spans biochemistry, science communication, bioethics and science policy. She has worked for a national bioethics advisory council, the Irish Council for Bioethics (2005-08), as policy adviser at the Wellcome Trust Sanger Institute (2008-13), and at the Francis Crick Institute (2013-14).

Stephanie joined McGill University in 2014, where she has been conducting ethics and policy research focusing on data sharing policy with large international collaborations, such as the Global Alliance for Genomics and Health (GA4GH) and the International Human Epigenome Consortium (IHEC). She spent 4 years at the Centre of Genomics and Policy in McGill's Department of Human Genetics and is now working with McGill's Montreal Neurological Institute on its Tanenbaum Open Science Institute and the Canadian Open Neuroscience Platform (CONP).

Stephanie co-chairs the GA4GH Researcher Identities Task Team and is a member of several other GA4GH Workstreams, CONP Ethics & Governance and Communications Committees, and IHEC Bioethics and Communications Workgroups. She also serves as a member of the NHLBI Observational Study Monitoring Board for the Myelodysplastic Syndromes Natural History Study. Previously, Stephanie served as Chair of McGill Epigenome Mapping Centre Data Access Committee (2016-18) and member of the International Cancer Genome Consortium Ethics and Policy Committee (2009-13).

Marc Fiume
DNAStack, Canada

Marc Fiume is an entrepreneur, researcher, and thought-leader in genomics. Earned his doctoral degree exploring the intersection of biology and computer science and is committed to developing transformational technologies to better quality and longevity of human life. Highly collaborative with multiple supervisory, mentorship, leadership, and teaching roles. PhD. training in collaboration with Dr. Stephen Scherer at Hospital for Sick Children, inventing novel software algorithms and applications for the detection of genetic mutations, particularly structural and copy number variations. Co-founder and CEO of DNAstack, Toronto-based genomics software company with a mandate of making genomics and clinical information globally accessible and useful, supported by major cloud providers, and academic and clinical research partners around the world. Strong presence supporting Toronto- and Canadian-led genomics initiatives and sequencing centres. Co-founder of the Canadian Genomics Cloud. Co-Chair of the Beacon Project for the Global Alliance for Genomics & Health, defining global technical, ethical, and security standards for genomic mutation sharing. Founder and Lead of the Beacon Network, the world's largest search engine for public sharing of genomic mutations, which has facilitated millions of searches and has led to disease-gene discoveries and clinical translation. Co-Chair of the Discovery Work Stream for the Global Alliance for Genomics & Health, with the mandate of building the Internet of Genomics, i.e. digital network infrastructure to connect genomics and clinical information systems to accelerate genomics and biomedical research and discovery worldwide.

Paul Flicek
EMBL-EBI, United Kingdom

Paul Flicek is a Team Leader and Senior Scientist of the European Molecular Biology Laboratory and leads the Vertebrate Genomics Team at the European Bioinformatics Institute (EMBL's Hinxton Outstation) near Cambridge, England. He is head of the collection of EBI resources focused on Genes, Genomes and Variation and in this role provides strategic leadership of the four faculty groups that provide all of EBI's genomics and variation databases and resources. He leads the Ensembl project and also has leadership roles in data management activities for the 1000 Genomes Project, the International Human Epigenome Consortium (IHEC) and the International Mouse Phenotyping Consortium (IMPC). Flicek's research is focused on comparative regulatory genomics and the evolution of transcriptional regulation. He is also interested in the large-scale infrastructure required for modern bioinformatics including storage and access methods for high throughput sequencing data. He has played a role in a number of major international projects including ENCODE and the mouse, chicken, and gorilla genome projects.

Robert Freimuth
Mayo Clinic, USA

Robert Freimuth, Ph.D., works at the interface of medical informatics, bioinformatics and genomics to develop computational methods, tools and infrastructure that will speed the translation of advances in genomics to clinical practice.

The long-term goal of Dr. Freimuth's research is to develop resources that make genome-guided therapy a routine part of clinical care.

Dr. Freimuth's research includes designing scalable and semantically interoperable systems that are based on standardized ontologies and terminologies, information models, and structured data elements. These systems are essential for integrating the large data sets and diverse knowledge bases that form the foundations of personalized medicine.

Dr. Freimuth is exploring methods for integrating genomic data into the Mayo Clinic electronic medical record (EMR) and developing clinical decision-support tools that enable physicians to understand and make use of a patient's unique genomic data. The initial focus of his work is in the area of cancer pharmacogenomics, currently one of the most promising applications of genome-guided therapy.

David Glazer
Verily, USA

David Glazer is an engineering director at Verily Life Sciences, where he helps life science organizations use cloud computing to accelerate and scale their work with big data. He is a PI for the Data and Research Center, and a member of the Steering Committee, of the NIH *All of Us* Research Program, and he serves on the NIH Advisory Committee to the Director. He is co-chair of the Cloud Workstream, and a member of the Steering Committee, of the Global Alliance for Genomics and Health (GA4GH). He previously worked at Google, where he founded the Google Genomics team, and led a variety of platform, product, and infrastructure teams. Prior to joining Google in 2006, he successfully started two companies: Eloquent in 1995 (IPO 2000), which used rich media to power business communications, and Verity in 1988 (IPO 1995), which did full-text search. David grew up in Massachusetts, where he earned a BS in physics from MIT.

Peter Goodhand
Global Alliance for Genomics and Health, Canada

Peter Goodhand is a leader in the global health sector as a senior executive and board member.

Goodhand played a key role in the creation of the Global Alliance for Genomics and Health (GA4GH) and was appointed as its founding Executive Director in 2014, and as CEO in 2018. Since May 2016, he has also served as the President of the Ontario Institute for Cancer Research (OICR).

Prior to the GA4GH and OICR, he was the President and CEO of the Canadian Cancer Society, Canada's largest health charity. Before joining the charitable sector, Goodhand had a 20 year career in the global medical technology industry, including strategic leadership roles with multinational healthcare companies such as American Cyanamid and Johnson & Johnson; Board Chair and President of Canada's Medical Device Industry association (MEDEC); and as the founding Managing Director and then Board Chair of the Health Technology Exchange (HTX).

He is currently Chair of the Steering Committee of the Occupational Cancer Research Center, Co-chair of the Medical and Scientific Advisory Board of Global Genes, board member of the AGE-WELL Network Center of Excellence, and on the Steering Committee of the Global Genomic Medicine Collaboration (G2MC).

He chaired the Government of Canada's Expert working group on the future of medical isotope production, and was a member of the Canadian delegation to the UN summit on non-communicable diseases.

Ian Green
SNOMED International, United Kingdom

Ian Green is based in the UK, and has worked for SNOMED International for the last 5 years in a variety of roles. Ian's current role covers Customer Relations management for Europe, leading Clinical Engagement activities and a particular focus on developing SNOMED CT's capacity to support Genomics developments globally.

He is a nurse by background, who moved into healthcare management in both primary and secondary care sectors, and then into Health Informatics. Previously, he worked for the NHS in the UK, authoring both READ codes and SNOMED CT for over 10 years. In addition to a general interest in all areas of health informatics, he has a particular interest in engaging with clinicians to develop a clinical terminology that meets the requirements of all clinicians globally that can transform the way healthcare is delivered for the benefit of patients. Ian obtained an MSc in Quality in Healthcare, focusing on the implementation of evidence-based medicine and clinical governance, with a particular emphasis on quality management processes.

Ian's interests are reading, music and motor racing, and spending time with his family.

Jean-Pierre Hubaux
EPFL, France

Jean-Pierre Hubaux is a full professor at EPFL. Through his research, he contributes to laying the foundations and developing the tools for protecting privacy in tomorrow's hyper-connected world. He has pioneered the areas of privacy and security in mobile/wireless networks and in genomics.

He is the academic director of the recently created Center for Digital Trust (C4DT). He leads the ETH-funded project Data Protection in Personalized Health (DPPH) . He held visiting positions at the IBM T.J. Watson Research Center and at UC Berkeley. He is one of the seven commissioners of the Swiss FCC and a member of the "Information Security Task Force", set up by the Swiss federal government. He is a Fellow of both IEEE (2008) and ACM (2010). He is among the most cited researchers in privacy protection and in information security.

Julius Jacobsen
Queen Mary University of London

Julius Jacobsen has previously worked at the EBI in the UniProt group developing software to automate the mapping between UniProt and PDB entries as part of the SIFTS project. He extended this to create a pipeline for automating the annotation of PDB entries in the UniProtKB/TrEMBL database and developed internal tools to help UniProt curators incorporate this data in UniProtKB/Swiss-Prot.

He spent a brief period of time in the finance industry as an software development engineer in a global payment card processing enterprise, but missed working in the biological domain. After this, he returned to the Wellcome Trust Sanger Institute to work with the International Mouse Phenotyping Consortium (IMPC), helping to produce the disease-gene association data in collaboration with other members of the Monarch Initiative.

He is currently lead developer of the Exomiser, a bioinformatics tool for analysing genomic data and prioritising candidate genes based on the patient phenotype. He has helped incorporate the Exomiser software into the 100,000 genomes project clinical pipeline. He also contributed to modules of the Matchbox software for matching rare-disease patients as part of the Match Maker Exchange (MME) project. Most recently, he has been working on Phenopackets - a driver project for the GA4GH Clinical Phenotypic Data Capture and Exchange Work Stream.

Peter Kapitein
Inspire2Live,
Netherlands

Peter Kapitein is a Patient Advocate of Inspire2Live. Peter connects patients, researchers and clinicians to further research, treatments and care; in the Netherlands as well as international. He organizes congresses, lobbies the matrix of public authorities, health care organizations, insurance companies and health research institutes. Peter also gives lectures and talks to help patients and society to fight cancer where possible and live with cancer with a good quality of life. He writes blogs, articles and books that contribute to these topics. Peter has studied the Medical Industrial Complex, the complex in which the stakeholders in healthcare work together in a way that does not necessarily benefit the patient. Health care is (without bad intention) distracted from its essence: the patient.

Peter is the co-founder of Alpe d'HuZes, the foundation that is most famous for the annual cycling event on Mount Alpe d'Huez and that raised over 150 million euro for the fight against cancer. He works at the Central Bank of the Netherlands as a program manager and advisor for complex and politically difficult problems. His employer facilitates him in this job. His job enables Peter to be genuinely independent and to work tirelessly for the interests of all patients globally. Peter was honoured with a doctorate in October 2012 at the Free University in Amsterdam for connecting patients, researchers and clinicians all over the world.

Thomas Keane
EMBL-EBI,
United Kingdom

Thomas Keane is a faculty member at EMBL-EBI and Team Leader for the European Genome Phenome Archive (EGA), the European Variation Archive (EVA), as well as the Archival Infrastructure for the European Nucleotide Archive (ENA). He is responsible for strategic planning for EGA, ENA (Archival Infrastructure) and EVA, is a member of the Global Alliance for Genomics and Health (GA4GH) steering committee and leads the Large-Scale Genomics workstream. He is the scientific lead for the Mouse Genomes Project, a collaboration with the Wellcome Sanger Institute.

Bron Kisler
NCI, USA

Bron Kisler is currently working with the US National Cancer Institute (NCI) as a Biomedical Informatics Specialist through Essex Management, where he is a Senior Associate. He primarily supports the Center for Biomedical Informatics & Information Technology (CBIIT) as part of the Semantic Infrastructure and Enterprise Vocabulary Services teams, providing expertise in data standards, semantics / controlled terminology, data models, metadata repositories as well as data science and informatics solutions. Mr. Kisler is applying his expertise across a number of "Big Data" programs under the Beau Biden Cancer Moonshot Initiative such as the Cancer Research Data Commons and Genomics Data Commons. Additionally, He is assisting key NCI programs with implementation of CDISC standards for data collection and regulatory submissions.

Mr. Kisler has 30 years of technical and business experience from both the public and private sectors. He has worked as an entrepreneur and innovator in the healthcare and clinical research marketplaces for over 20 years. Mr. Kisler is a Founder of the Clinical Data Interchange Standards Consortium (CDISC) and served as Vice President, Strategic Alliances & Development. He was also a Director of the CDISC Europe Foundation. As a Founder of CDISC and long-time member of the Executive Team, Mr. Kisler played a pivotal role in CDISC's growth and international recognition with membership spanning nearly 30 countries. CDISC standards have been widely implemented around the world and are required for clinical trial submissions in the US and Japan. He has served as Primary Investigator on FDA Grant projects for Lung Cancer, Prostate Cancer, Colorectal Cancer, Cardiovascular Endpoints, Schizophrenia and Virology.

He holds 3 Bachelor of Science degrees in Mathematics, Computer Science and Statistics.

Ilkka Lappalainen
CSC - IT Center for Science, United Kingdom

Ilkka Lappalainen is the Deputy Head of ELIXIR Finland and leads a group that provides data management services for the national research organisations. The aim is to ensure secure, scalable and international standards compliant research environment through active engagement with organisations such as ELIXIR, BBMRI and EuroBioImaging. Ilkka works closely within the GA4GH Discovery Stream and coordinates the Finnish involvement in many other works streams such as DURI, Cloud, Large Scale Genomics, Clinical&Phenotypic Data Capture. Before his current position he worked as the ELIXIR Human data coordinator at the ELIXIR-Hub and a Project lead for the EMBL-EBI European Genome-phenome Archive (EGA) and Database of Genomic Variations archive (DGVa). He gained postdoctoral experience at the University of Cambridge and a PhD from the University of Helsinki focusing on biophysical and bioinformatical properties of genomic variants causing rare disorders.

Peter Lichter
German Cancer Research Center (DKFZ), Germany

Peter Lichter is head of the Division “Molecular Genetics” at the German Cancer Research Center (DKFZ), Heidelberg since 1992, Co-director of the National Center for Tumor Diseas (NCT), Heidelberg since 2015, Editor-in-chief of the International Journal of Cancer since 2009 and Speaker of the Heidelberger Center for Personalized Oncology (DKFZ-HIPO) since 2011. Upon graduating from the University of Heidelberg, he trained as a postdoctoral fellow at the Yale University Medical School with Prof. Dr. David C. Ward. In 1990 he was appointed as a junior group leader at the DKFZ, before he became head of division. Peter Lichter has made significant contributions in the areas of molecular cytogenetics, pioneering protocols for FISH and array-CGH, tumor pathomechanisms and architecture of the cell nucleus. In recent years he has made major contributions to the field of translational genome research in oncology.

Mike Lin
DNAnexus, USA

Mike Lin is a researcher and software developer in genome informatics, whose work enables numerous large-scale genome projects in collaboration with DNAnexus, GA4GH, and other organizations. Previously, he led genomics R&D and cloud systems engineering at DNAnexus, after studying computational comparative genomics at MIT and the Broad Institute.

Chris Lunt
NIH, USA

Chris Lunt is the CTO of the All of Us Research Program, an endeavor to collect comprehensive health information, including a full genome, for one million Americans. This information will be used to transform precision medicine. Chris has been a technology executive for nearly twenty years, having previously worked on public/private health insurance systems, crowd work, digital identity and social networks, and information ontologies. He is co-inventor of a dozen seminal social networking patents. He has started multiple companies, and has one IPO. He is an avid musician, writer, and reader.

Michaela Mayrhofer
BBMRI-ERIC, Austria

Michaela Th. Mayrhofer earned her PhD from both the Ecole des Hautes Etudes en Sciences Sociales and the University of Vienna (shortlisted by the Austrian Society for Political Science for 'best thesis 2010'). She was investigator in several national and international research projects focusing on the politics and governance of the life sciences. Her academic career led to various positions at the Centre de Recherche Médecine, Sciences, Santé et Société, the University of Vienna, the Institute of Science, Technology and Society Studies at Alpen-Adria-Universität Klagenfurt/ Vienna/Graz, the Technical University of Vienna and the Medical University of Graz. Today, she serves as BBMRI-ERIC's Chief Coordination & Policy Officer, contributes to the coordination and knowledge generation of the Common Service ELSI, leads the ELSI Work Packages for ADOPT BBMRI-ERIC and CORBEL, and leads the Code of Conduct for Health Research initiative.

Olivier Menzel
BLACKSWAN Foundation, Switzerland

Olivier Menzel graduated (B.Sc.) from the University of Geneva where he obtained a Master of Medical Genetics (M.Sc.) in 2001 and a PhD in 2006 from the University of Lausanne and EPFL at the Swiss Institute for Experimental Cancer Research (ISREC). For seven years he directed the laboratory of pediatric surgery at the University Hospital of Geneva. In parallel he created the BLACKSWAN Foundation (blackswanfoundation.ch), a Swiss foundation to support research on rare and orphan diseases, organized international scientific conferences (RE(ACT) Congress; react-congress.org) and launched an online platform for sharing scientific knowledge and crowdfunding (RE(ACT) Community; react-community.org). For two years he was a director of a company specialized in the identification, acquisition, development, marketing and sale of research programs for rare and orphan diseases. In 2013 he obtained an Executive MBA from the HEC of Lausanne with a specialization in Management Healthcare. He served as director at the second largest group of private clinics in Switzerland, Swiss Medical Network, then, for 2 years he was the Managing Director and Consultant atThink Rare Sàrl and fully involved in the BLACKSWAN Foundation activities. Now he is the Head of Strategic Partnerships at the Health 2030 Swiss Genome Center.

Anna Middleton
Wellcome Genome Campus, United Kingdom

Anna Middleton is a creative social sciences researcher with expertise in designing innovative surveys and conducting focus groups to explore and understand what people think about the application of genomics. She is passionate about delivering research that has real practical value. She has been the Founder and Head of the Society and Ethics Research group at the Wellcome Genome Campus since October 2016. She is also a Professor/Affiliate Lecturer, Faculty of Education, University of Cambridge.

Anna is involved in the genetic counselling profession internationally and is current Chair of the Association of Genetic Nurses and Counsellors. She co-wrote and co-leads two modules of the University of Cambridge MSt Genomic Medicine course and co-created the first World Congress on Genetic Counselling.

Her current research focuses on: 1) international lay public attitudes towards anonymised DNA data being a part of Big Data (Your DNA, Your Say), 2) use of films in the NHS with patients to explain genomics as part of genetic counselling (Music of Life) and 3) a collaboration with Theatre of Debate to explore public attitudes toward the expectation of doctors to share relatives' genetic results with others.

Madeleine Murtagh
Newcastle University, United Kingdom

Madeleine Murtagh is Professor of Sociology and Bioethics at the Policy Ethics and Life Sciences Research Centre (PEALS), Newcastle University, UK. Madeleine's early research examined how people interact with and make decisions about using new health treatments and technologies, later expanding to consider practices of (open) data science in health (particularly in longitudinal cohort studies and biobanks) and health data ethics. Madeleine conducts social studies of sociotechnical and normative practices in health research data sharing and governance as well as developing new forms of governance which centrally involve citizens and research participants in ethical and governance decision-making. This work is carried out in the context of transdisciplinary and intersectoral research teams, often in interventionist, collaborative ethnographic studies. Madeleine is co-lead of the Regulatory and Ethics Work Stream of GA4GH.

Hidewaki Nakagawa
Japan Agency for Medical Research and Development (AMED), Japan

Hidewaki Nakagawa graduated from Osaka University School of Medicine in 1991, and he completed training in clinical oncology of GI & breast cancers and critical care medicine as a surgeon. After obtaining his PhD from Osaka University for hereditary GI cancer genomics, he had postdoctoral training (1999-2003) of Lynch syndrome and cancer genomics at the Human Cancer Genetics Program, The Ohio State University, USA. In 2003, he returned to Japan as an assistant/associate professor at Institute of Medical Science, The University of Tokyo, where he was dedicated to therapeutic target screening for cancers. In 2008, he moved to RIKEN as a team leader of genomic medicine and his recent research focuses on cancer genomics by whole genome sequencing and GWAS, cancer immunology, and biomarker development using NGS. He has been working for cancer genome sequencing projects of liver and GI cancers as one of PIs of international collaborative ICGC and PCAWG. Since 2015, he has been also dedicated as a program officer of Biobank/Genomic Medicine project in Japanese medical funding agent, AMED (Japan Agency for Medical Research and Development), for genome research and genomic medicine promotion in Japan and Asia.

Kathryn North
Murdoch Children's Research Institute, Australia

Kathryn North is Director of the Murdoch Children's Research Institute and the David Danks Professor of Child Health Research at the University of Melbourne.

Professor North is trained as a physician, neurologist and clinical geneticist and was awarded a doctorate for research in neurogenetics. She completed a postdoctoral fellowship in the Harvard Genetics Program.

Professor North is a national and international leader in Genomic medicine. In 2014, she was appointed as Vice Chair of the Global Alliance for Genomics and Health – a collaborative network of over 500 organisations across over 70 countries. Commencing in 2016, she leads an NHMRC-funded national network of over 80 institutions - the Australian Genomics Health Alliance.

Professor North has received a number of awards including the GSK Australia Award for Research Excellence (2011), the Ramaciotti Medal for Excellence in Biomedical Research (2012) and Member of the Order of Australia (AM) for service to medicine in the field of neuromuscular and neurogenetics research (2012). She chaired NHMRC Research Committee (2012-2018) and International Advisory Board of the Great Ormond Street Institute of Child Health (UK) and is a member of the Board of the Victorian Comprehensive Cancer Centre.

Corrie Painter
Broad Institute, USA

Corrie Painter is the associate director of operations and scientific outreach in the Cancer Program of the Broad Institute of MIT and Harvard where she oversees the development, launch and operations of several patient-partnered genomics studies where patients can consent online to donate their stored tumor samples, saliva samples, blood samples medical records, and their voices in order to directly accelerate the pace of discovery (https://www.broadinstitute.org/count-me-in). As part of these efforts, she leads the Angiosarcoma Project, aimed at generating the genomic landscape of this orphan disease.

Prior to joining the Broad Institute in 2015, Painter was vice president and cofounder of Angiosarcoma Awareness Inc., a nonprofit devoted to fostering a collaborative atmosphere between researchers in order to generate data and reagents that can be shared by the sarcoma community as a whole. She continues in this role alongside her work at the Broad Institute.

Heidi Rehm
Broad Institutel, USA

Heidi L. Rehm is the Chief Genomics Officer in the Department of Medicine and at the Center for Genomic Medicine at Massachusetts General Hospital. She is also Medical Director of the Broad Institute Clinical Research Sequencing Platform and Professor of Pathology at MGH, Brigham & Women's Hospital and Harvard Medical School. She is a board-certified laboratory geneticist and leader in defining standards for the interpretation of sequence variants and a principal investigator of ClinGen, providing free and publicly accessible resources to support the interpretation of genes and variants. Rehm also co-leads the Broad Center for Mendelian Genomics with Daniel MacArthur focused on discovering novel rare disease genes and co-leads the Matchmaker Exchange to aid in gene discovery. She is a strong advocate and pioneer of open science and data sharing, working to extend these approaches through her role on the steering committee of the Global Alliance for Genomics and Health. Rehm is also a co-investigator of the BabySeq Project exploring the clinical use of genomic sequencing as an adjunct to newborn screening, principal investigator in the eMERGE consortium supporting genomic discovery and genomic medicine implementation, and a principal investigator on a project to develop i2b2 into a Health Innovation Platform for clinical decision support.

Augusto Rendon
Genomics England, United Kingdom

Augusto Rendon obtained an undergraduate degree in Physic (magna cum laude) at the Universidad de los Andes, Bogota, Colombia. Augusto completed his doctoral degree at the University of Toronto in the Department of Medical Biophysics. He then moved to the University of Cambridge to pursue a postdoctoral position in computational biology. Augusto currently holds a Principal Research Associate position at the University of Cambridge in the Department of Haematology. His research has focused on understanding the relationship between genotype and phenotype in human blood formation, using a wide range of angles including epigenetics, transcriptomics, GWAS, and rare disease genetics.

In June 2014 Augusto was seconded to Genomics England as Director of Bioinformatics for the UK's 100,000 Genomes Program. There, he leads a team of about 50 developers, analysts, bioinformaticians and curators. His team is responsible for establishing the pipelines to analyse and manage all genomic data for the project. They have designed and implemented the clinical interpretation pipelines that feedback findings to patients, while ensuring that knowledge accumulated through this process is best exploited to improve patient care and enhance discovery. His team also develops databases to store hundreds of millions of variants for hundreds of thousands of patients.

Gary Saunders
ELIXIR, United Kingdom

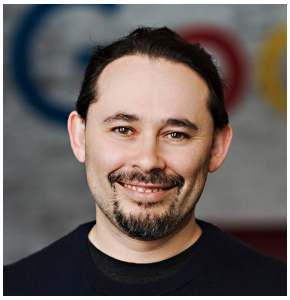
Gary Saunders is Human Data Coordinator at ELIXIR. He leads the implementation of the ELIXIR-wide strategy to enable responsible sharing of human data consented for reuse in scientific research. A major focus of this role is working with the existing Human Data Communities to ensure that all data generated is compliant with the FAIR data principles, and to coordinate these efforts with the Global Alliance for Genomics and Health (GA4GH).

Previous to ELIXIR, Gary was at EMBL-EBI where he was the data manager for the European Variation Archive (EVA), and the Database of Genomics Variants Archive (DGVa). He has a PhD in Bioinformatics from the University of Glasgow, UK, and has a background in comparative genomics.

Serena Scollen
ELIXIR, United Kingdom

Serena Scollen is the Head of Human Genomics and Translational Data at ELIXIR, the European infrastructure for bioinformatics and life-science data, based in Hinxton, UK.

Prior to joining ELIXIR, she was a Director within the Human Genetics group at Pfizer. In this role, she led and implemented a genetic and precision medicine strategy to support drug target selection and clinical programmes for the Pain and Sensory Disorders Research Unit. She was also a member of the ABPI Stratified Medicine Working Group. Earlier in her career, she worked within the Toxicogenomics group at GlaxoSmithKline. She gained postdoctoral experience at University of Cambridge and Imperial College London and a PhD from the University of Cambridge, with a focus on the genetic susceptibility to disease.



Ilia Tulchinsky
Google, Canada

Ilia Tulchinsky is a software developer with over 20 years of experience spanning medical devices and integrations in neurology and electrodiagnostic fields as well as mobile, media and data mining. In his 10 years at Google, Ilia has built scaled applications and infrastructure across enterprise applications, mobile and information retrieval systems and contributed to Cloud Genomics platform and open source efforts.

Ilia is the Engineering Director within Google Cloud Healthcare and Life Sciences, focusing on large scale Cloud based computation support for healthcare protocols, data formats and APIs and enabling secure and compliant high performance analytics workflows and machine learning in the Cloud.

Peter Van Reusel
CDISC, Belgium

Peter Van Reusel is a data standards expert with a passion for implementing practical solutions that work. He started his career in a large pharma company as a database analyst and has been working with data standards all his career.

In 2008, he started the Business & Decision Life Sciences CRO, specialized in CDISC Conversions and Biometric Services. He assumed the role of Chief Operating Officer, responsible for group operations, QA and IT.

Peter is a certified CDISC CDASH and SDTM trainer, delivering courses across Europe and he is the past chair of the CDISC E3C committee.

Since June 2018, Peter is the Chief Standards Officer for CDISC where he is responsible for the development and execution of the CDISC Data Standards strategy.

Alex Wagner
Washington University School of Medicine, USA

Alex Wagner is currently a postdoctoral research fellow in the Griffith Laboratory at Washington University School of Medicine. His graduate research in the Braun Laboratory at the University of Iowa focused on the identification of germline genomic variants associated with rare heritable retinal dystrophies. There, Alex developed the Ocular Tissue Database of tissue-specific gene expression (genome.uiowa.edu/otdb), developed a machine learning model for prioritizing retinal disease variant candidates, and used integrative RNA-seq and exome sequencing to discover non-exomic and synonymous mutations linked to Stargardt disease. Since his postdoctoral work began in 2015, he has co-developed several web tools to assist in the interpretation of somatic genomic variants in cancers, including the Drug-Gene Interaction Database (DGIdb; dgidb.org), the Clinical Interpretations of Variants in Cancers knowledgebase (CIViC; civicdb.org), and the Database of Curated Mutations (DoCM; docm.info). He also led the analysis of a relapsed small cell lung cancer cohort, discovering the role of WNT signaling activation as a mechanism of acquired chemoresistance in this disease. Alex has more recently been working with the Variant Interpretation for Cancer Consortium (cancervariants.org), a driver project of the GA4GH, to harmonize the content of CIViC and other variant interpretation knowledgebases to improve variant interpretation consensus.

Andrew Yates
EMBL-EBI, United Kingdom

Andrew Yates is a Team Leader at the European Molecular Biology Laboratory's European Bioinformatics Institute (EMBL-EBI); near Cambridge, UK. He has worked in bioinformatics for 16 years and provides interface development, data access APIs, production support, outreach and training for the Ensembl (http://www.ensembl.org) and Ensembl Genomes (http://www.ensemblgenomes.org) projects. Ensembl is one of the primary methods researchers access human genome annotation in a free and open source environment. Andrew currently co-leads the Genomic Knowledge Standards GA4GH work stream, which develop, adopt, and adapt standards-based components to enable the exchange of reference genomic information through common APIs and enabling the interpretation of genomic data within a clinical setting. He also co-leads the refget specification, a component of Large Scale Genomics, which enables access to reference genomic sequences using a decentralised identifier system.

Andy was educated at the University of Manchester gaining a BSc(Hons) in Pharmacology and a MSc in Bioinfomatics before joining the Cancer Genome Project at the Wellcome Genome Campus, UK.

Rumiana Yotova
University of Cambridge,
United Kingdom

Rumiana Yotova is a Lecturer and Director of Studies in Law at Gonville and Caius College, University of Cambridge and an Affiliated Lecturer at the Faculty of Law. She practices as a Door Tenant at Thomas More Chambers. Rumiana teaches and does research in the areas of public international law and EU law. She is particularly interested in international biomedical law and recently advised the Nuffield Council on Bioethics on the regulation of heritable human genome editing under international, comparative and EU law.

EUROPEAN
GENOME-PHENOME
ARCHIVE

10th Anniversary Symposium

October 5

16:00 - 21:00

Singapore Hall

Celebrate 10 years of the European Genome-phenome Archive!

AGENDA

16:00

Welcome and Introduction

Jordi Rambla (CRG)

16:15

Sharing human data: why, how and what can we achieve?

Serena Scollen (ELIXIR Europe)

16:45

Challenges and opportunities in data sharing infrastructure in Africa

Nicola Mulder (University of Cape Town & H3Africa)

17:45

The EGA: 10 years of Ethical Genomic Access

Adrian Thorogood (McGill University)

18:15

The future of EGA in a world of federated human data

Thomas Keane (EMBL-EBI)

Registration Required | Drinks & Canapés to follow

Hosted by EMBL-EBI

Location & Venue

Congress Center Basel
Messepl. 21, 4058
Basel, Switzerland

The Congress Center Basel is situated downtown and directly adjacent to Basel Exhibition Centre. Travel times are short, and the historic old town, the shopping areas and the many museums and parks are walking distance or a few minutes away by public transport. The Congress Center Basel and its facilities are located directly alongside Messe Exhibition.

Download the conference center floor plan:

- 2nd Floor
- 3rd Floor

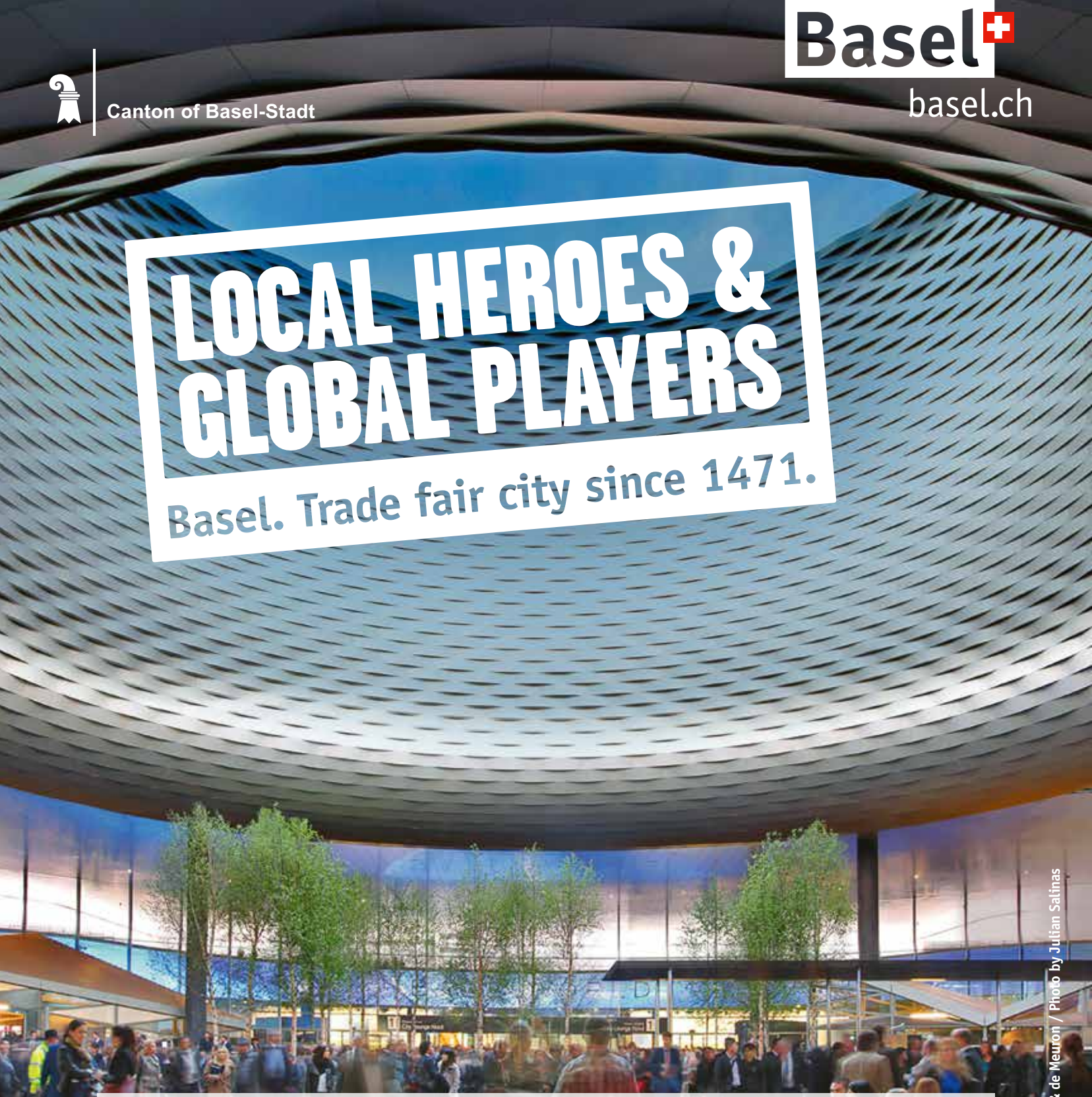
Travel by Plane	The Congress Center Basel is a 15 minute drive from the Basel-Mulhouse-Freiburg Airport and is accessible by bus (Bus Nr. 50), tram (Line No. 2 from Bahnhof SBB), or taxi.
Travel by Railway	All three of Basel's railway stations — SBB (Switzerland), SNCF (France) and DB (Badischer Bahnhof, Germany) — are within 5 to 10 minutes drive to the Congress Center Basel.
Travel by Car	Messe Basel and the trade-fair grounds have their own motorway exit. The "Messe" exit from the A2 motorway leads directly to the fair and congress ground. There is a multi-storey car park at Exhibition Square. From there, follow the signs directing you to the Congress Center Basel.

Canton of Basel-Stadt

Basel
basel.ch

LOCAL HEROES & GLOBAL PLAYERS

Basel. Trade fair city since 1471.



City Lounge of the Messe Basel by Herzog & de Meuron / Photo by Julian Salinas

For hundreds of years, Basel has been an attractive location for successful trade fairs, congresses and major international events. The Basel Autumn Fair, for example, which dates back to 1471, is the oldest and biggest funfair in Switzerland. And Baselworld, global trade fair for watches and jewellery, as well as Art Basel, the most important art fair in the world, attract major international players and numerous visitors every year. You'll be very welcome!

www.basel.ch CityBasel

Dining and Local Attractions

Whether you're looking for an open-air art tour, a sightseeing tour in a panorama bus, or excitement for water fans or animal lovers — with the diversity of activities on offer in Basel you're spoilt for choice.

Basel Tourism has put together a list of top activities you can do during your stay at Switzerland's cultural capital.

- **Take a trip across the Rhine in style and comfort on one of the four ferries that connect Kleinbasel with Grossbasel.** Between Basel's five bridges across the Rhine you will find the city's four ferries, "Wilde Maa", "Leu", "Vogel Gryff" and "Ueli", with which you can cross the Rhine without motorised assistance, using only the natural power of the river's current.
- **Walk through Basel's Old Town.** There can be few other cityscapes where buildings dating back as far as the fifteenth century engage so harmoniously and vibrantly with the contemporary creations of internationally distinguished architects. Tickets for a guided walking tour are available online.
- **Visit the museums of Basel**. As Switzerland's city of museums, Basel attracts visitors with internationally renowned galleries such as Fondation Beyeler, Kunstmuseum (Museum of Fine Arts) and Museum Tinguely. There are close to 40 museums to admire in Basel.
- **Enjoy the food!** The city's unique location guarantees you a varied and top-quality range of culinary delights. Visit the Basel Tourism website to find local restaurants that will suit your need.

Visit the Basel Tourism website (https://www.basel.com/en) for more information on dining and local attractions in Basel.

Open call for new GA4GH Driver Projects

These real-world genomic data initiatives help guide our development efforts and pilot our tools. We will accept up to 5 new Driver Projects in 2019 which will be announced in early 2019 and will meet as many of the following criteria as possible:

Driver Project Criteria

- ✓ Global representation
- ✓ Scientific merit
- ✓ Capacity to contribute

Visit **ga4gh.org** for more details and to apply