THE ASSOCIATION OF GENETIC NURSES AND COUNSELLORS

SPRING MEETING

Programme and Abstract Booklet

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Cancer Research UK Cambridge Research Institute
Li Ka Shing Centre
Robinson Way
CAMBRIDGE
CB2 0RE

(P11) Sharing genomic research data: launch of new study

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Elements of a person's past, present and future medical health can now be revealed in a matter weeks via whole exome sequencing of a saliva or blood sample. Such technology is frequently used in research to understand the genomic basis of disease and will very soon be used within clinical health services.

It has been considered good practice for many years to conduct genomic research anonymously and not share any individual results with research participants. However, there is mounting pressure to change this approach and begin to share individual results with research participants. There is, however, no clear guidance or evidence to suggest what sort of data research participants want - should it only relate to clinically actionable conditions or would participants be interested in receiving results with broader implications or even their raw sequence data? There is an urgent need for large-scale empirical research to gather evidence on what is reasonable, responsible and indeed ethical to share.

As a contribution to this process we have designed a mixed-methods study and have launched an innovative questionnaire (see www.genomethics.org) that uses film to explore the ethical implications of whole genome research. The questionnaire is accessed online. We are inviting genetic counsellors, genomic researchers, health professionals and lay members of the public world-wide to complete the questionnaire and are aiming for 20,000+ responses. This poster will introduce the study design, explain why the research into genome ethics is important and discuss the relevance of this to both the research and clinical genetics community.

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(P12) Recurrence risk and germ cell mosaicism in X- linked adrenoleukodystrophy

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Recurrence of X-linked conditions in subsequent children has been observed in families where parents are not constitutional mutation carriers. The usual explanation
for this is germ cell mosaicism. There is very little information in the literature about germ cell mosaicism in X-linked adrenoleukodystrophy (ALD). The symptoms of ALD vary widely but onset of the severe form usually begins in boys before age 10 years and is characterised by a rapid cerebral demyelination, increasing disability and death. Around 95% of affected boys have inherited the causative ABCD1 mutation from their mother. This case study focuses on the difficulties of quantifying the recurrence risks of ALD for a mother who is not a constitutional carrier of the ABCD1 mutation identified in her son. Possible recurrence of ALD in the sibling of a boy with an apparently de novo mutation is discussed in the context of what is known about germ cell mosaicism in other X-linked conditions. Other issues raised by this case with implications for practice include: explaining germ cell mosaicism, decision making around prenatal diagnosis and ALD carrier testing in healthy sisters of an affected boy with an apparently de novo ABCD1 mutation.

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