Most people eager to know the secrets of their genetics

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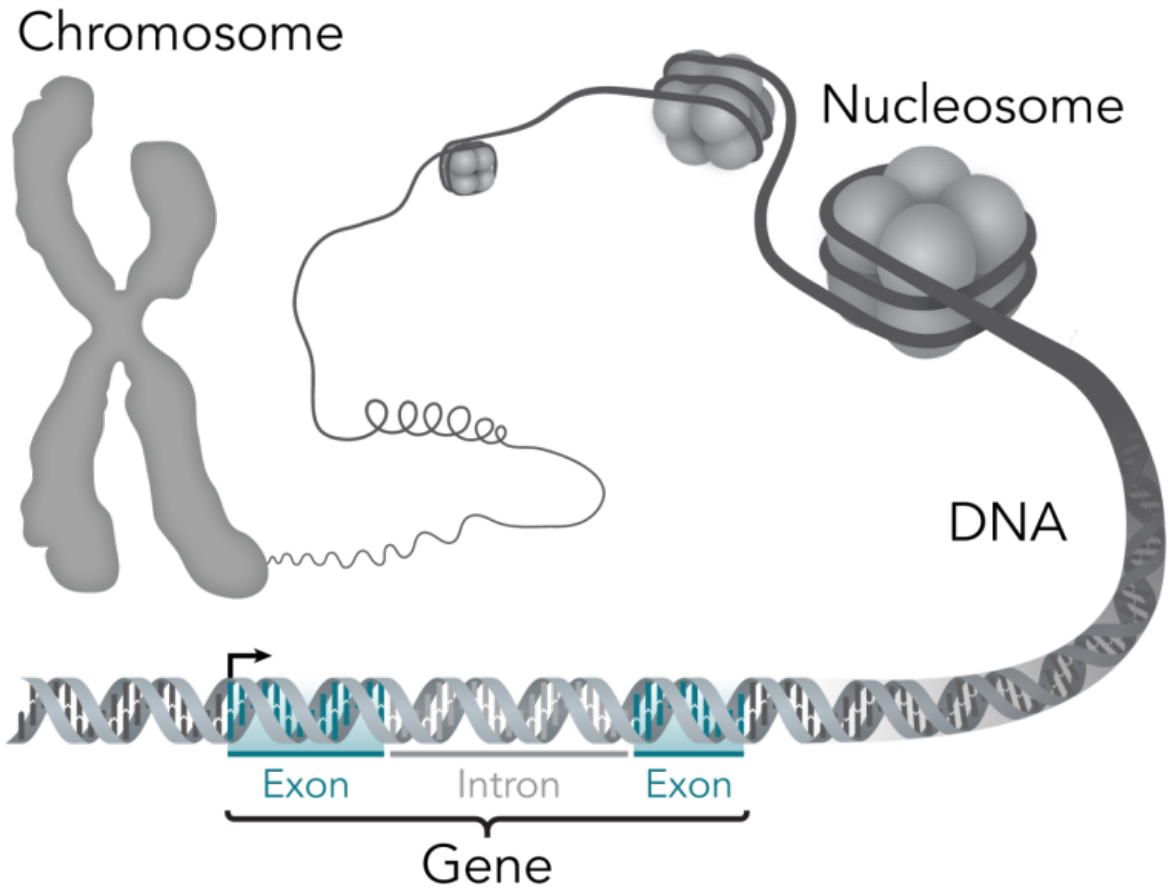

This stylistic diagram shows a gene in relation to the double helix structure of DNA and to a chromosome (right). The chromosome is X-shaped because it is dividing. Introns are regions often found in eukaryote genes that are removed in the splicing process (after the DNA is transcribed into RNA): Only the exons encode the protein. The diagram labels a region of only 55 or so bases as a gene. In reality, most genes are hundreds of times longer. Credit: Thomas

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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.

The results show that genomic data has a perceived value to participants even if it is not currently clear what the information means for health outcomes. However, in general, the majority of people were interested in clinically actionable data and genetic professionals surveyed were concerned about returning data that cannot yet be interpreted accurately.

"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."

The usefulness of findings was an important consideration for the majority of respondents, who were drawn from 75 countries around the world. The majority of participants wanted to receive information about serious conditions, even if the risk of developing the condition was as low as 1 per cent. However, fewer people were interested in receiving results for less serious conditions.

"When we asked patients and families how much they want to know about their genetic information their immediate reaction was that whatever information the researchers or clinicians found out, they wanted to know too," says Alastair Kent OBE, Director of Genetic Alliance UK, a charity that works to improve the lives of people affected by genetic disorders. "But there can be no one size fits all. We need to make sure that there is enough information and support available to allow individuals to make an informed choice about what is right for their situation. We need to remember this information belongs to the individual and they should be able to decide for themselves what they do and don't find out about their health - which means we need to start thinking about how this can be recognised."

Of the interest groups surveyed, genetic health professionals were five times more likely than other groups to think that incidental findings, results that are not the main focus of a research project but may be of clinical importance, should not be shared. Both genetic health professionals and genomic researchers were more likely to think that information about ancestry should not be shared.

"Genetic health professionals are acutely aware of the challenges posed by interpreting genetic information accurately and communicating results to patients," explains Dr Helen Firth, an author from the Department of Clinical Genetics at Addenbrooke's Hospital. "There are still so many unknowns; having key indicators for a disease in your genetic code may not necessarily mean that you will develop that disease. Much of the information in our personal genetic codes is currently uninterpretable and of uncertain clinical significance. It will take many years of research before we know how to use much of this data for clinical benefit. As this knowledge is gained, our survey will help researchers and health policy makers to plan accordingly."

The survey was conducted as part of the Deciphering Developmental

Disorders (DDD) project, which seeks to find genetic diagnoses for rare developmental disorders using patients' sequence data. The DDD project did not search this data for disease indicators and only returned results likely to be linked to the patients' disorders. This fits well with the findings of the survey as, while participants were keen to learn about their genetics, the majority did not think researchers should be required to actively search for key indicators of disease in genomic data if it would reduce the time and resources spent on medical research.

More information: Middleton, A et al. (2015). Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research. *European Journal Human Genetics*. DOI: 10.1038/ejhg.2015.58

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