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Experts urge caution over use of new genetic sequencing techniques

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The use of genome-wide analysis (GWA), where the entirety of an individual's DNA is examined to look for the genomic mutations or variants which can cause health problems is a massively useful technology for diagnosing disease. However, it can also pose major ethical problems if used incorrectly, say new recommendations from the European Society of Human Genetics (ESHG) published on line today (16 May 2013) in the *European Journal of Human Genetics*.

Many services based on whole genome and on exome* sequencing and analysis are now available to patients at an affordable price, and this raises the question of how to ensure that they are provided appropriately. "Such sequencing generates huge amounts of information that needs to be processed, analysed, and stored in a responsible manner", said Professor Martina Cornel, chair of the Professional and Public Policy Committee of ESHG. "It is preferable to use sequencing or analysis specifically targeted at a particular health problem to avoid unsolicited findings, or those that cannot yet be interpreted, which can cause considerable anxiety to patients and their families. Clear guidance on how to deal with such findings is needed."

Targeted analysis will limit such unsolicited findings, says the ESHG, and this is particularly important at present when there are only a limited number of clinicians properly trained to inform patients on the significance of the results of GWAs and exome sequencing. While the Society believes that the duty to inform patients may outweigh their right not to know in some circumstances, the new recommendations propose

that analysis should be limited to genome regions linked to the clinical problem for which the analysis is being undertaken.

"We are opposed to the type of opportunistic screening that throws up large numbers of incidental results. If such results reveal a treatable or preventable condition, then clearly it is advantageous to patients to be informed about them. But in the majority of cases it is very difficult to interpret exactly what such incidental results mean for patients and their families. The evidence currently available often comes from families with affected persons, but it is lacking on the interpretation of results in other situations. Furthermore, in genetics healthcare, autonomy is considered very important: patients should be allowed consent on what would be screened for and reported to them. We believe that it is premature today to look for such results other than the clinical problem in circumstances where there are no prior clinical indications or family history ", said Professor Cornel.

"A sustained effort to educate clinicians in genetics is needed in order to be able to cope with advances in analysis. We also believe that the Society has an important role to play in raising awareness of genetics among the general public. Only with the benefit of a general increase in genetic literacy can society become properly involved in the debate over who has the right to know what and in which circumstances," she said.

Professor GertJan van Ommen, Editor in Chief of the *European Journal of Human Genetics*, said: "The importance of this issue has been underlined by the US Government's Bioethics Advisory Panel's plans to report on how incidental findings encountered in genomics research should be handled. I believe that ESHG has made an important contribution to the debate, which will be further discussed at their conference in Paris in June."

*Exomes are the short sequences of DNA representing the regions in

genes that are translated into protein

More information: Whole genome sequencing in human health care: Recommendations of the European Society of Human Genetics Volume 21, Issue S1 (June 2013). www.nature.com/ejhg/journal/v21/n1s/index.html

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