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Should the results of individual genetic studies be disclosed to participants?

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Individual results of genetic research studies should not be disclosed to participants without careful consideration, a scientist will tell the annual conference of the European Society of Human Genetics today (Monday). Dr. Robin Hayeems, from the Department of Health Policy, Management and Evaluation at the University of Toronto, Canada, will say that she believes that the view held by many ethicists that individual genetic research findings should always be reported to participants involved in genetic research studies was perhaps misguided, and that to do so without careful consideration of evidentiary assumptions and clinical capacity could distort the responsibilities of researchers and lead to misunderstanding.

Dr. Hayeems leads the GE3LS (Genomics and its Ethical, Economic, Environmental, Legal and Social Aspects) component of a Genome Canada funded basic science project that is looking at identifying the genes that can modify the severity or clinical effects of cystic fibrosis (CF). Together with the study co-lead Professor Fiona Miller, in charge of the GE3LS component of the Genome Canada funded autism genome project, her team surveyed researchers from around the world who were involved in genetics research related to CF and autism. "We were interested in their perspectives about sharing genetic research results with individual study participants in order to be able to add their voices to the ongoing debate about whether and under what circumstances researchers are under an obligation to report these results to research participants", she said.

The survey found that 80% of the researchers agreed that individuals in whom a genetic variation had been identified should be informed of this finding if it were judged to be clinically significant. Yet it also revealed considerable variation among researchers in deciding when a result was clinically significant. Researchers felt less confident about the clinical significance of a result when the finding was related to autism research, was less scientifically robust, and was incidental to the condition being studied. Further, researchers were 40% less likely to report it when they were unable to provide participants with the requisite medical advice related to the finding. There were also differences between scientific disciplines, with clinical researchers being 1.8 times more likely to class a particular finding as clinically significant and 1.5 times more likely to report it to study participants than were molecular and statistical researchers.

"Our understanding of how genetic factors contribute to the heterogeneous collection of conditions that comprise the autism spectrum disorders is in its infancy. By contrast, though much remains to be learned about the genetics of CF, the clinical consequences of classical CF and the basic genetic defect that causes it has been known for some time", said Dr. Hayeems.

"I think our discovery that an autism-relevant finding engenders less confidence with respect to clinical significance compared to a CF-relevant finding reflects researchers' implicit sense of the fundamental uncertainty that still prevails with respect to the genetics of autism. What is interesting about the survey design is that we can say overall that confidence in an autism-related genetic finding was lacking compared to a CF-related finding, even when the autism-related finding they were asked to judge was, by design, quite robust.

"Most, but not all, ethicists endorse an obligation to report genetic research results about individuals because they consider it to be clinically

relevant information that individuals have a right to receive. This argument presumes that these research results constitute such information, and that the judgment of clinical significance is relatively straightforward. Our work suggests that this presumption may be misplaced. The results of the survey identify a set of factors that appear to influence researchers as they consider whether a result is clinically significant and whether it should indeed be reported. These factors go beyond scientific standards of robustness to include underlying uncertainties about the role of genetics in certain conditions, as well as researcher training and research team capacity", she said.

The GE3LS team now intends to encourage institutional research bodies and the wider research ethics community to revisit their thinking about the obligation to report research results to include a broader set of factors so that the complexity of the issue is fully reflected. "Our work highlights an important intersection between health research and health care", said Professor Miller. "This intersection raises important questions. Are results being interpreted and reported in the context of a research relationship in which the norms of clinical care cannot be expected or, in the context of clinical care, in which case these norms are assumed? What context is assumed, and who is responsible - researchers, or health care systems - for ensuring appropriate disclosure and follow up?" she concluded.

Provided by European Society of Human Genetics