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DTC genetic tests neither accurate in their predictions nor beneficial to individuals

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Amsterdam, The Netherlands: Direct-to-consumer (DTC) genetic tests give inaccurate predictions of disease risks and many European geneticists believe that some of them should be banned, the annual conference of the European Society of Human Genetics will hear today (Tuesday). In the first of two studies to be presented, Rachel Kalf, from the department of epidemiology at Erasmus University Medical Centre, Rotterdam, The Netherlands, will say that her research is the first to look at the real predictive ability of such tests, the results of which are available directly to an individual without having to go through a healthcare professional.

Working under the supervision of Associate Professor Cecile Janssens, together with researchers from Leiden, The Netherlands, and Boston, USA, Ms Kalf examined the risk predictions supplied by two large DTC companies, deCODEme (Iceland) and 23andMe (USA). They simulated genotype data for 100,000 individuals based on established genotype frequencies and then used the formulas and risk data provided by the companies to obtain predicted risks for eight common multi-factorial diseases – age-related macular degeneration (AMD), atrial fibrillation, celiac disease, Crohn's disease, heart attack, prostate cancer, and Type 1 and Type 2 diabetes (T2D).

Although the predictive ability of the DTC tests in the study was moderate for all diseases, both companies assigned an increased risk to a substantial part of the group. Yet the risk of disease in this group was often not substantially higher than the risk in the rest of the population

studied. For AMD, the disease with the highest predictive ability, both companies assumed that the risk in the population was around 8%. Of all subjects designated as having an increased risk, 16% using the 23andMe risk estimations and 19% using deCODEme's estimations would develop AMD, compared to the 4% found in the rest of the population studied.

"So individuals in the increased risk group may have a four-fold increased risk of disease, but they are still far more likely not to develop the disease at all. For T2D, where the companies calculated the average risk at around 25%, 32% of those assigned to the increased risk group would actually develop T2D compared to 22% in rest of the study population. This difference in disease risk is too small to be of relevance", said Professor Janssens.

"deCODEme predicted risks higher than 100% for five out of the eight diseases", Ms Kalf will say. "This in itself should be enough to raise considerable concern about the accuracy of these predictions – a risk can never be higher than 100%. In the case of AMD one in every 200 individuals in the group would have received a predicted risk of higher than 100%, suggesting that they would definitely develop the disease."

The DTC companies have been criticised for giving an exaggerated and inaccurate message about the connection between genetic information and disease risk. "They only take genetic factors into account when predicting risks for consumers, whereas in most multi-factorial diseases other modifiable risk factors, such as diet, environment, exercise and smoking have a much stronger impact on disease risk", said Professor Janssens. "We are all aware of the ethical problems surrounding DTC genetic testing, but this study also confirms that their predictions are inaccurate. At a time when some governments are considering regulating such tests, we believe that we have made an important contribution to the debate", she concluded.

In the second study, Dr. Heidi Howard from the University of Leuven, Belgium, and her colleague Professor Pascal Borry reported the results of a survey of a representative sample of clinical geneticists from 28 countries across Europe on their experience of and attitudes to DTC genetic testing. "This is the first ever survey of European clinical geneticists on the subject", Dr. Howard will say, "and the results were conclusive - 69% of respondents felt that prenatal gender tests should be legally banned, and 63% wanted to proscribe whole genome scans carried out by DTC companies."

One of the problems with DTC tests is that the companies' tendency to overstate the potential of predictive information does not help to produce a public properly educated about the potential value and limitations of genetic information. This is particularly true when it comes to whole genome scans, where a lot of results are given for many different conditions, the researchers say.

"Clinical geneticists' concerns with DTC genetic tests are mostly related to the fact that these tests usually lack clinical validity and utility. Moreover, these tests are usually carried out without the provision of genetic counselling. According to the experiences of clinical geneticists, patients often do not know how to interpret the results they receive and are often confused by them. However, almost all clinical geneticists feel that they have a duty to provide counselling if patients contact them after having purchased a DTC genetic test," says Dr. Howard.

"A person who undergoes a genetic test has to be accompanied – explanations, physical aid, the right to choose whether to know or not – and this is not true in the case of direct access to such a test", said one survey respondent.

"Genome-wide scans by companies are totally unacceptable, as they can derive sensitive information about medically relevant conditions and will

also provide lots of information which is difficult to interpret, even for medical professionals", said another respondent. Presenting the results of such tests directly to individuals is unacceptable, the majority of those surveyed said.

90% of respondents felt that a pre-symptomatic test - predicting if an asymptomatic person had a very high probability of developing a condition - should not be allowed without face-to-face medical supervision; 93% felt the same for a predictive test for a condition that has a penetrance (the proportion of individuals with the mutation who exhibit clinical symptoms) of 50 – 60%; 79% for a carrier test for homozygous monogenic disorders, such as sickle-cell anaemia; and 72% for a predictive test for a condition that increased or decreased a person's risk of developing it by 4% when compared to the general population.

At the moment, DTC genetic tests reach the market without having undergone any form of regulation. "Better regulation is needed at the level of market introduction of these tests", says Professor Borry. As in the case of drugs, a procedure should be developed for genetic tests."

Currently only a few European countries, for example France and Switzerland, have legislation that states that genetic tests can only be accessed via individual medical supervision. "Although this model is sometimes criticised for being too paternalistic", says Professor Borry, "in the absence of a good working pre-market control of genetic tests, it could be a useful way of responding to some of the concerns over DTC testing."

The provision of genetic testing services outside the regular healthcare system can also lead to unnecessary visits to healthcare providers and hence an increased burden on healthcare resources, the researchers say.