

Interpreting DTC testing results imposes a major burden on genetics services

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The increasing popularity of direct to consumer (DTC) genetic testing is having an impact on clinical genetics services, according to Australian researchers who will present their work to the 53rd annual conference of the European Society of Human Genetics, being held entirely on-line due to the COVID-19 pandemic, today [Saturday]. Many consumers are unsure about what to do with the results they receive, and many general practitioners are ill-equipped to advise them, meaning that they turn to clinical genetics services for help.

Ms Jane Tiller, Ethical, Legal and Social Adviser in Public Health Genomics, Monash University, Victoria, Australia, and colleagues analysed how often clinical genetics services were receiving referrals related to DTC testing, and what actions were taken by the clinic after receiving the referral. Until now, this subject has received little attention. "We knew that clinical genetics services had limited resources and long waiting lists. We wanted to explore the impact of DTC-related referrals in order to be able to forecast effects on the delivery of clinical genetics services and inform policymakers so that they could adjust resource needs accordingly," she says.

The researchers surveyed eleven publicly-funded Australian clinical genetics services, asking questions related to the DTC-generated referrals they had received over the past ten years. They found that 83% of such referrals were made by [general practitioners](#), in order to aid interpretation of results, and that over 30% of referrals were related to imputed disease risk estimates, where an online tool interprets raw

genetic data to obtain a health risk profile. The services reported that DTCGT results were often unreliable; fewer than 10% of the results tested were validated.

Currently the national regulator, the Therapeutic Goods Administration (TGA) is considering the issue of DTC genetic testing. Because the sale of health-related DTC testing is prohibited in Australia, some Australians obtain this testing through overseas companies or use data from ancestry sites to obtain imputed health data.

"If the TGA relaxes these restrictions on DTC genetic testing, this could increase yet further the need for clinical genetics services by consumers of DTC tests," says Ms Tiller. "Our study raises many issues. There are tensions between the desire to allow consumers access to their genetic information and subsequently managing and funding the healthcare follow-up that they need, either to interpret their results, or to determine an individual risk management plan. Commercial companies who gain from selling these tests often neither consider nor contribute to the cost in downstream health management."

In addition to the need for sufficient resources and funding for public genetics services, measures that could be taken to improve the situation include increasing the genomic knowledge of general practitioners. "GP knowledge ranges from the extremely genomically literate to the extremely genomically illiterate. There is no way of guaranteeing that all GPs have a sufficient degree of genomic literacy to interpret potentially inaccurate DTC results. Improving genomic literacy across GPs would certainly assist with this aim, and is critical as genomic testing becomes more common," she says.

Another problem that needs addressing is the lack of a consistent policy for referral management across Australia. Some services provide an appointment to all DTC-related referrals, some provide none, and some

assess referrals on a case-by-case basis, leading to considerable inequalities in access. A uniform national policy regarding the treatment of DTCGT-related referrals should be developed to reduce inequality and ensure consistent decision-making by publicly-funded genetics services in this area.

Now we need to look in further detail at year on year rates of referral, rates of validation and specific disease risk. As DTC [genetic testing](#) continues to grow, we expect its impact on health services will increase. Although our study was carried out in Australia, our findings are relevant for other countries with publicly-funded or single-payer clinical genetics. Policymakers need to take note and act now in order to facilitate efficient operation of clinical genetics and to provide the maximum benefit to the population," Ms Tiller will conclude.

Chair of the ESHG conference, Professor Joris Veltman, Dean of the Biosciences Institute at Newcastle University, Newcastle upon Tyne, UK, said: "Direct to consumer (DTC) genetics tests are becoming more widely available in society and this study asks the question whether this increases the workload for clinical genetics services in Australia. Overall, my impression is that the number of referrals related to these DTC tests is rather low, but it does make clear that most GPs do not know what to do with these and that shows a need for more education in this area."

Provided by European Society of Human Genetics

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