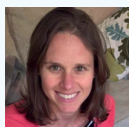




GENETIC COUNSELLING

Verity Leach looks at the genetic counsellor's role, offering support to families dealing with the impact of genetic diagnoses in the fast-changing, emerging field of genetic medicine.



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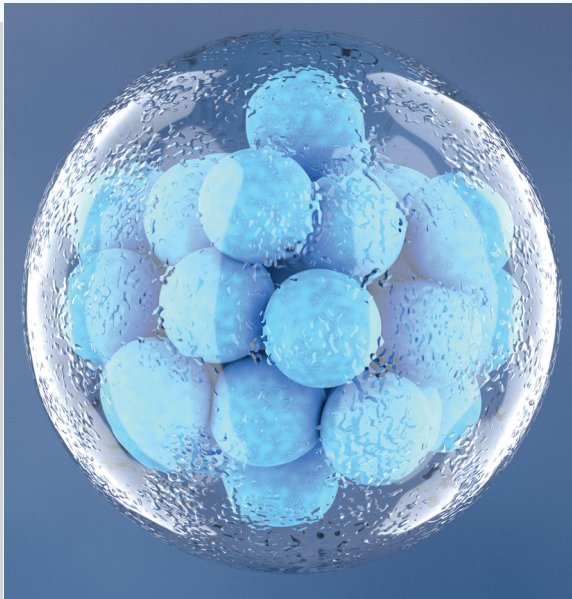
Few people have heard of the role of a genetic counsellor (GC), but those who have, know us well. A GC works closely with patients and families, offering genomic information, analysing family histories, ordering and interpreting genomic test results, and offering support in light of those results. The title, therefore, is a little misleading (since it does not involve therapeutic counselling). But we do use counselling techniques to help patients adjust to having a genetic condition and to help them make difficult decisions associated with this. We work

closely with multidisciplinary teams, including consultant geneticists and clinical scientists, as well as staff from other disciplines, such as obstetrics, cardiology, and paediatrics.

My faith provides me with a grounding in my job by giving me some ethical parameters within which I feel comfortable operating. It also helps me – to some extent – make sense of the suffering our patients often face. Sometimes I find, however, that my faith is at odds with the information and options I present to my patients – particularly in prenatal work. Those with a previously known condition sometimes choose IVF, where only unaffected

key points

- Genetic counselling is a relatively new field that uses counselling skills to help patients and their families adjust to the diagnosis of a genetic disorder.
- Much in the field is changing rapidly, and what can now be tested for was unknowable just a few years ago. But this creates new problems for families.
- The author explores how her faith helps her support and guide families through the confusion of data, options, tests, and the implications of a diagnosis.



◀ embryos are returned. Many genetic conditions are tested for during pregnancy, varying from early to late gestation. Following testing, parents often face difficult decisions.

Initially, I found providing information about ending pregnancies – some of them very late term – difficult. I have come to realise, however, that my job is to provide information and support. Ultimately, it is the parents who are responsible for their decisions. At the same time, I always provide the important option that it is okay to carry on with the pregnancy.

As medicine progresses and we are able to offer so much more in genetics – even compared to five years ago – it can feel as though the testing odyssey never ends. We can and do provide our patients with a lot more understanding and answers. But with that comes expectations that are hard to meet. Patients want answers; they want to know why a certain situation has happened to them. Even when genetics can provide answers, the effects can ripple down families for generations – especially with the more complex genomic conditions – which can make offering support difficult. The role of a GC is to manage expectations and provide realism, including making it clear that any genomic finding will impact not only that one nuclear family but also their much wider family network.

As a Christian, I sometimes wonder whether it is better not to open the Pandora's box that is genetic testing, particularly as our rate of incidental findings is becoming more significant as technology improves. In the end, our faulty genes are only an expression of the sin that has dislocated everything good in God's creation. And even when we can find the cause of a genetic condition, it does not

necessarily solve the problem. We need a better saviour than genetic testing for that! The Bible makes it clear that we don't always have the answers. We need to know a contentment that only comes from Jesus.¹ The constant striving for answers indicates that some patients are trying to take some control in an often uncontrollable situation. Sometimes, I find it more helpful to ask, 'how can I help you live with uncertainty and a lack of knowledge?' rather than, 'let me see what further testing we can offer'.

The area of practice I find most challenging to my faith is the question all of us who work in healthcare know: why do some people have so much pain and sadness in their lives whilst others never experience such grief? This could be the family who have been affected by early onset Huntington's disease in every generation (where the uncertainty of inheriting the condition only compounds the grief and guilt for many families). It might be those who experience the loss of multiple children or who have several children with severe disabilities. Then there is the family marred by a young cancer diagnosis. It can feel desperate at times, and I often find myself

burdened by sadness. As a Christian, I have faith in the Lord Jesus. I know my final destination, and this provides me with a sure and certain hope. But many of my patients endure these difficult situations without this faith and yet cope with such grace and strength. I often wonder if I would respond similarly even though I have a relationship with the living God who loves me.

The areas of my practice most at odds with my Christian faith have probably not fully arrived yet. As medicine progresses – particularly genetics – and the values of our

society become less rooted in the Judaeo-Christian worldview, I wonder at what point I will have to reassess my practice. Advanced genetic testing for children (pre or post-birth), the implications of which could lead to parents being able to 'design' their baby, transgender issues, and private testing options are all ethical concerns that could provide challenges in the future. I am unsure what these challenges will mean for the role of a GC, but as Christians, we are called to obey the Word of the Lord, which is never changing, and apply it to a medical and ethical landscape that is ever-changing. ◦

Even when genetics can provide answers, the effects can ripple down families for generations



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reference

1. Philippians 4:11