

For your family's future

Offering couples the invaluable service of genetic carrier testing, Genetic Clinics Australia helps couples make informed reproductive choices about their future family's health. Genetic counsellor **Nikki Gelfand** tells us more.

Q: What are the types of genetic screenings available at Genetic Clinics Australia?

Nikki Gelfand: Genetic Clinics Australia is currently focusing on genetic carrier testing before pregnancy, called preconception carrier screening. These tests check if couples are carriers of genetic conditions which could affect their future children. Recent technological advances in testing means that testing can be done for just one, a few or over 100 conditions at once. Although these tests are ideally performed before pregnancy, they can also be performed early in pregnancy. We also organise genetic screening for conditions like Down Syndrome, which are usually performed in the first trimester of pregnancy.

Q: Who are people who should consider genetic testing? Are there certain ethnicities more susceptible to genetic conditions?

NG: We believe all couples starting a family, or having more children, should consider this type of genetic testing prior to pregnancy. The American College of Obstetricians and Gynaecologists (ACOG), and the Royal Australian and New Zealand College of Obstetrics and Gynaecology (RANZCOG) both have statements recommending this type of testing be offered to all couples, preferably prior to pregnancy. One in four Caucasians are carriers of a serious genetic condition and have no family history of the condition to make them aware of it. Many of these conditions are extremely serious and have a devastating impact on the child and their families.

Different ethnic groups are more susceptible to certain genetic conditions, which differ around the globe. Some examples include thalassaemia in people of Middle Eastern and



South East Asian origins and Tay Sachs Disease in the Ashkenazi Jewish population.

Compared to the wider population, individuals of the same ethnic group come from a much smaller gene pool, with less genetic variation. This means a couple could actually be distantly related as they could both be descendants of a common ancestor, especially if their history involved migration.

If this common ancestor carried a genetic fault for one of these conditions, the chance of these two people being carriers of the same genetic change is increased. This is called a "founder effect", i.e. caused by a founding member of that branch of the population. When marrying within an ethnic group is favoured, it can then further increase the frequency of these genetic changes.

Interestingly, it has recently been shown that the distinctions of ethnicity are becoming less relevant due to migration and intermarriage in both present and past generations. As a result, using ethnicity as a guide to determining testing does not necessarily exclude individuals from being carriers of many genetic conditions. For example, a person may appear African American, and yet have many Caucasian genes and vice versa.

For this reason, we believe that couples from

all ethnic groups would benefit from being tested for many of the more common recessive genetic conditions, regardless of family history or ethnicity.

Q: How are results from genetics testing helpful to a couple?

NG: Results of pre-conception carrier testing for genetic conditions gives couples valuable information about genetic risks for their future family planning. Couples with this information can be empowered to consider various reproductive options to avoid having children born with these serious genetic diseases.

For example, when this testing is done before pregnancy, the option of using a technology called Pre-implantation Genetic Diagnosis (PGD) is available. PGD involves genetic testing of embryos created through In-Vitro Fertilisation (IVF). Healthy embryos can then be selected, avoiding the genetic condition in the resulting baby.

Alternatively, some couples elect to get pregnant naturally, and undergo testing during pregnancy, called prenatal diagnosis, for the genetic condition in question.

There are pros and cons to the various different options that become available to couples in this situation.

Additionally, since we all receive our genetic information from our parents, results of genetic testing can also be beneficial to other family members, such as siblings and cousins.

Q: What is the role of a genetic counsellor at Genetic Clinics Australia?

NG: Genetic counsellors are health professionals, with extensive training in both genetics and counselling. Our role is to help individuals and couples understand complex genetic information and support them through often extremely difficult decision making.

Genetic counsellors practice in a non-directive manner, meaning we will not tell anyone what to do, but rather present all the options in a non-biased way and then support people to make decisions consistent with their own beliefs and values.

Genetic counsellors also help families to cope with the burden of living with genetic conditions, and support family members to share important genetic information with their relatives. Our role varies according to the needs of different patients in different situations. Of course, confidentiality is an extremely important aspect of a genetic counsellor's work as well.

Q: Is pre-conception carrier screening supported by rabbis and other religious leaders?

NG: We find that many of the community's rabbis and other religious leaders are supportive of genetic testing prior to pregnancy, and many rabbis recommend this testing to their congregants prior to marriage.

This testing can ensure parents are spared the heartache of an unexpected diagnosis of a serious condition in their child.

Many religious leaders support utilisation of technologies such as PGD and prenatal diagnosis, particularly if their use can reduce the suffering of the family.

Sadly, many families in our community understand first hand the reality of living with these types of conditions, as this type of genetic testing has not been widely available in the past. Genetic carrier screening is now easier than ever to access, and more accurate and detailed than ever before.

Genetic Clinics Australia is staffed by two experienced genetic counsellors, Nikki Gelfand and Andrea Leibler, and medical director Dr Jonathan Cohen. For more information, visit www.geneticclinic.com.au or call (03) 9528 1910 to book a confidential appointment.