Reproductive carrier screening

For many years, we have been able to offer screening for a small range of conditions in pregnancy that occur if there is a change in the usual DNA pattern. The most common of these is Down Syndrome, known medically as trisomy 21, and many women undertake screening for this in their pregnancy. It is also important for pregnancy caregivers to ask about any inherited conditions that are known to occur in a family, as these can also impact on the health of a new baby.

There are many other DNA changes that can affect the health of a baby. Our DNA contains small sequences called 'genes' which are the templates used to make the proteins from which we are put together. An error in a gene is called a 'mutation' and if this change affects how the protein works, disease can occur in the person. Some diseases only occur if a person inherits a damaged copy (or mutation) of the same gene from both parents – this is called a 'recessive' mutation. In these diseases, there is often no history in the family, so the child might be the first person affected. The most well-known of this type of genetic disease is a severe and chronic lung disease called cystic fibrosis (CF). Some diseases of this type can be fatal to children at a very young age.

It is now possible to offer screening to couples planning pregnancy to see if they carry a mutation in a recessive gene that could affect a future baby. This is called reproductive carrier screening. Ideally this is done prior to pregnancy, but it can also happen once a woman is pregnant, although this limits the options available to the couple if a mutation is found. While many people may carry a mutation, it is important to remember that a recessive disease will occur only if both parents have a mutation in the same gene; this is not common.

If you are interested in discussing this type of screening further, please let us know and we would be happy to answer your questions. There is also information available on the internet, some of which is published by the companies offering these tests. In time, this type of screening may be available free of charge to all couples; this is the subject of a large trial starting later this year. However, at the moment, the laboratories that carry out the testing charge a cost starting at around $300 that is not covered by Medicare or private health insurance.

Please find below some of the company websites:
https://www.vcgs.org.au/tests/prepair
https://www.invitae.com/en/individuals/reproductive-genetic-testing/
https://www.sonicgenetics.com.au/rcs/
https://www.clinicallabs.com.au/patient/our-tests/pregnancy-testing/gene-access-carrier-screen/