

Generation® NIPT Genetic Counselling Service

Genomic Diagnostics is pleased to announce access to a new genetic counselling service. The service, provided in conjunction with myDNA Life is available to those patients who receive a high-risk result from their Generation® or Generation® Plus NIPT test (only for high-risk T21, T18, T13 and sex chromosome (X,Y) aneuploidy detected results). This is a high quality, free telephone service designed to enable patients to spend time talking to an expert genetic counsellor to answer any questions they may have about the implications of their test results. Further information about the genetic counsellors involved in this service are found overleaf.

Healthcare Practitioner FAQ's

1. When should a healthcare practitioner consider referring a patient to the Generation® NIPT Genetic Counselling Service?

The Generation® NIPT Genetic Counselling Service is designed to enable patients to discuss any questions they may have about the Generation® and Generation® Plus tests and to explore the issues and implications of their result from this test. The Generation® NIPT Genetic Counselling Service is only open to patients who have received a high-risk Trisomy 21, 18, 13 or sex chromosome (X,Y) aneuploidy result from either the Generation or Generation Plus NIPT test. The doctor should refer any eligible patient that would benefit from talking to a genetic counsellor.

Please note: genetic counselling for microdeletion or other chromosomal abnormalities is not covered by this service.

2. How is a patient referred to the Generation® NIPT Genetic Counselling Service?

Once the doctor has disclosed the result to the patient, they can refer their patient to the Generation® NIPT Genetic Counselling Service using the specific referral form provided by Genomic Diagnostics at the time of result notification. If the referral form has been misplaced, please call Genomic Diagnostics on 1800 822 999 or email info@genomicdiagnostics.com.au. The referral form must be fully completed and faxed to the Generation® NIPT Genetic Counselling Service number found on the referral form.

It is essential that the patient's current telephone contact details are written on the referral form.

3. When will the service be available?

The Generation® NIPT Genetic Counselling Service will be available from 9am-5pm, Monday to Friday AEST. A myDNA Life genetic counsellor will contact patients directly to arrange an appropriate appointment time.

4. What are the response times once a referral form has been submitted to the Generation® NIPT Genetic Counselling Service?

Initial contact with patients will be made by a myDNA Life genetic counsellor within 48 hours of receiving a completed referral form. The genetic counsellor will book an appointment time with the patient or if the timing is suitable, may discuss the situation immediately with the patient.

5. How will the Generation® NIPT Genetic Counselling Service be made aware of the patient's results?

Once myDNA Life have received the patient referral form, they will contact the Genomic Diagnostics customer care team and request a copy of the patient's results. This will be used as a reference during the consult. Doctors can also send a copy of the results with the referral.

6. What level of counselling will be provided to the patient?

The Generation® NIPT Genetic Counselling Service will make every effort to answer all questions raised by patients regarding their respective test result.

7. What happens after a patient receives genetic counselling via the Generation® NIPT Genetic Counselling Service?

Once the genetic counselling consultation is complete, the Generation® NIPT Genetic Counselling Service will:

- » Send the patient back to their referring healthcare practitioner for ongoing management and to arrange follow-up diagnostic testing (CVS and amniocentesis). As a small percentage of results will be false-positives, **follow-up diagnostic testing is the only way to get certainty and clarity about a high-risk result.**
- » Fax the referring healthcare practitioner a summary of counselling recommendations. Fax is being used to ensure efficient speed for communication.
- » Following the conclusion of the telephone consultation, myDNA Life will have no subsequent duty of care to the patient as they will be sent back to their referring healthcare practitioner.

8. Does the Generation® NIPT Genetic Counselling Service include counselling for Generation® Plus microdeletion or other chromosomal abnormality results?

The Generation® NIPT Genetic Counselling service is limited only to those who receive a high-risk result for Trisomy 21, 18, 13 or sex chromosome (X,Y) aneuploidy, and is **not** available to those who receive a high-risk result for the following:

- » A high-risk microdeletion result
- » A high-risk chromosomal abnormality result (other than Trisomy 21, 18, 13 or sex chromosome (X,Y) aneuploidy)

Any questions about the best option in such cases can be answered by calling Genomic Diagnostics on 1800 822 999.

9. Can a patient be referred to the Generation® NIPT Genetic Counselling Service for genetic counselling regarding other genetic conditions?

The NIPT Genetic Counselling Service is designed to enable patients to discuss any questions and explore any issues and implications of their high-risk Generation® or Generation® Plus NIPT result only.

The service is not designed to offer genetic counselling regarding other genetic conditions. If your patient has a family history of a genetic condition or is concerned about a genetic condition, please consider referring them urgently to a State Based Genetic Counselling Service (www.hgsa.org.au/asgc/find-a-genetic-counsellor).

The Team Supporting the Generation® Genetic Counselling Service



The Generation® NIPT Genetic Counselling Service is provided by myDNA Life, an Australian company that specialises in genetics and personalised medicine. Led by Associate Professor Leslie Sheffield, an experienced Clinical Geneticist, the team is staffed with experienced genetic counsellors who have many years of experience in both public and private healthcare sectors, working with many different genetic conditions.



A/Prof Leslie Sheffield

MB BS, MSc, FRACP, HGSA Cert Clin Genet

Since the 1980s Professor Sheffield has been involved in the design and development of genetic tests. As a Clinical Geneticist based at Royal Women's and Children's Hospitals, he authored more than 100 scientific publications. In 2007 Les founded myDNA to make genetic interpretation more widely available. He continues to drive the development of new services as Medical Director. He has been on the Editorial Board of Nature's Journal of Pharmacogenomics since 2006 and is currently the genetic editor of the Internal Medical Journal.



Vicki Petrou

Genetic Counsellor

FHGSA, Grad Dip Gen Couns, BAppSci MLS

Vicki is a certified genetic counsellor, who also has a strong background in laboratory science and quality and compliance. She spent a decade working in genetic counselling at the Victorian Clinical Genetics Service. She has mainly worked as a genetic counsellor on many different genetic screening programs. Vicki is cognisant of the importance of the provision of genetic counselling for patients that undergo screening tests. Vicki has also worked as a research genetic counsellor and is passionate about new developments in the field of genetics and the important role that genetic counsellors have in these areas.



Emma Harrison

Genetic Counsellor

MHGSA, BA, Post Grad Dip Arts (research) Bioethics, MGC

Emma is an associate genetic counsellor who is passionate about genetic education and increasing accessibility to genetic testing. Emma spent four years working in prenatal and preconception genetic counselling in Sydney and has worked closely with patients who have undergone non-invasive prenatal testing (NIPT). Since relocating back to Melbourne, Emma has become passionate about pharmacogenomics and commenced working as a genetic counsellor and health care professional liaison at myDNA.



Edith Sheffield

Genetic Counsellor

BA, Dip Ed, Grad Dip Gen Couns

Edith is a trained educator and genetic counsellor, who has experience in prenatal genetic counselling. Edith directed the education program at GenesFX Health (now myDNA Life) and provides genetic counselling input to the drug specific reports and to patients when required, as well as providing support for Doctors.