

Genetic Carrier Screening in General Practice

By Dr Caroline Rogers



Dr Caroline Rogers has been a GP on Sydney's Northern Beaches for the last 20 years and has been working at South Steyne Medical Centre in Manly since 2021. Her practice focuses on women's health and chronic disease, with a special interest in proactive, preventative care.

As GPs, we are ideally placed to discuss genetic carrier screening with patients. Over the last few years, I have started offering this test as part of my routine pre-conceptual and pregnancy planning consultations. When discussing carrier screening with patients, I often use the analogy of rhesus status. Just like rhesus testing, the screening is done once and

has implications for pregnancy care. Knowing about it sooner rather than later allows us to put plans in place to manage and prevent pregnancy complications. It is only necessary to test the partner if the patient tests positive.

The advantage of obtaining the results of genetic carrier screening before a pregnancy begins is that it reduces the stress and anxiety which can be associated with waiting for results while pregnant. If the test is positive for one or more genetic mutations and the partner also needs to be tested, the wait can be weeks. If this process is only initiated at the initial antenatal visit, we are often well into the second trimester before the patient knows for certain what the outcome is. This can significantly impact the joy and well-being many couples hope and expect to experience during this time.

Pre-pregnancy testing allows this to happen in a much less time-sensitive and emotionally charged environment. The patient and their partner can be referred to a genetic counsellor to discuss the nature of the conditions being screened for, if necessary, and their options in terms of pregnancy planning. This includes discussing preimplantation testing if a positive result is found for both parties.

As this is a once-in-a-lifetime test, and once cost is no longer a barrier, I anticipate it being raised more often during pill checks and cervical screening appointments with younger women who are not actively considering a pregnancy but would like to have this information to help them plan future fertility choices.

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