

Pre-Pregnancy and Early Pregnancy Genetic Screening



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Pre-pregnancy care is critical

“Pre-pregnancy counselling should include information about **carrier screening**... ideally pre-conception...”

- RACGP Guidance on Prenatal Testing

“Carrier status screening of women ... for the more common genetic conditions may be offered. Women ... should be appropriately informed of the benefits and limitations of testing... **pre-pregnancy screening is preferable to antenatal screening** for inheritable genetic conditions as this allows more options for carrier couples.”

- RANZCOG Genetic Screening Guidelines

“Every ... health care provider should establish a **standard approach to carrier screening** that’s consistently discussed with each patient and takes into account a patient’s family history and personal values. **All women should also be offered screening.**”

- ACOG

Preconception genetic screening involves taking a family history and pedigree and performing a risk assessment for pregnancy based on maternal age, family history and ethnicity.

If you are planning a pregnancy, genetic testing is available to identify whether you are at increased risk of having a child with a genetic condition.

Many healthy people are unaware they are carriers of a genetic disorder until they have an affected child. Improved technology means carrier testing can be undertaken for an increasing number of genetic conditions from a single blood or saliva sample. Testing is ideally performed prior to pregnancy to allow time for couples to consider all options available to them in accordance with their values and beliefs. Such conditions include; cystic fibrosis, spinal muscular atrophy, fragile X syndrome, as well as conditions common to different ethnic populations.

At Capital Genomic we can discuss the different preconception screening options available and facilitate testing appropriate to you.



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