Reproductive Genetic Carrier Screening

We would like to tell you about newly available reproductive genetic carrier screening, and implications of the screening and testing timeframes in pregnancy.

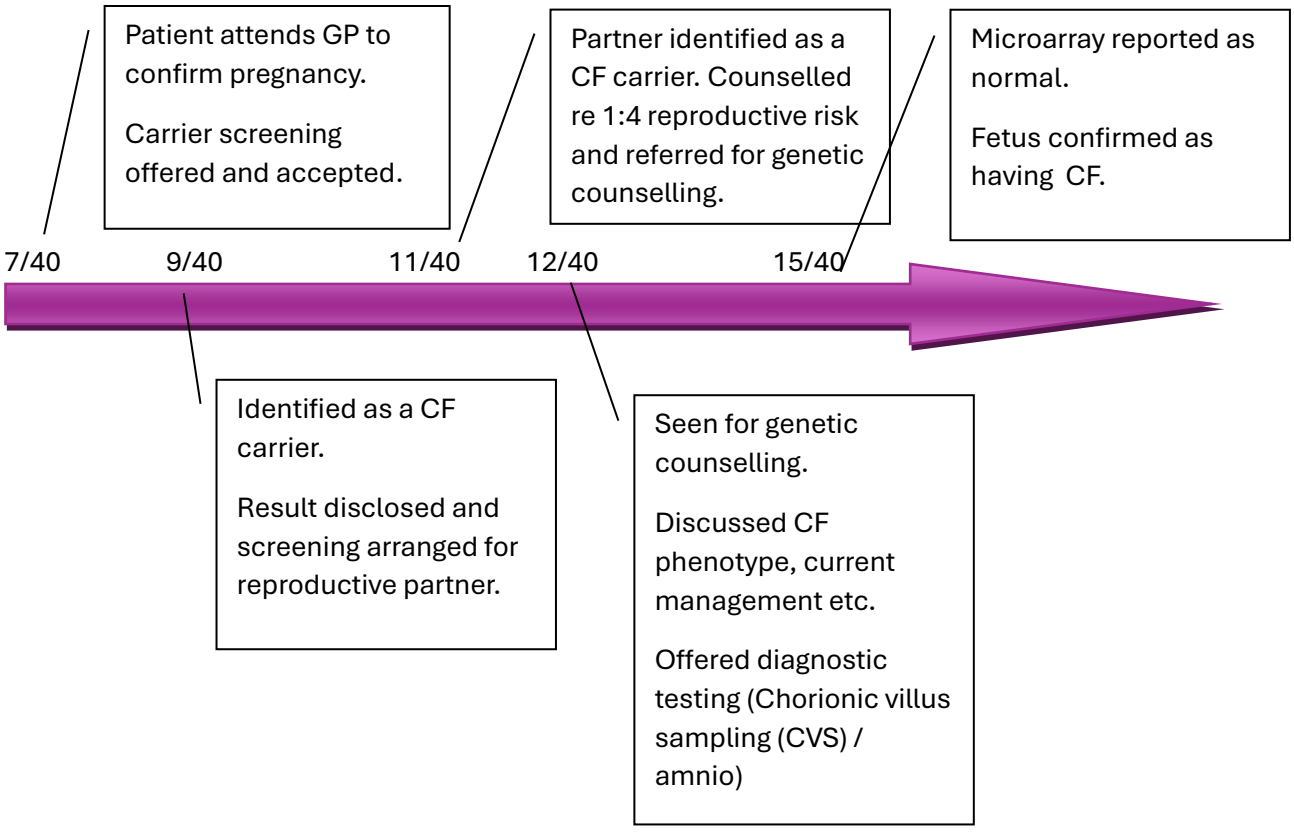
A new Medicare item number has been introduced for **reproductive genetic carrier screening**. This gives women the option to have screening for **three conditions**- cystic fibrosis (CF), spinal muscular atrophy (SMA) and Fragile X syndrome. Carriers are typically asymptomatic but at risk of having an affected child.

Male partners only can access Medicare funded testing if their partner has been identified as a carrier. Because of the timeframe of testing, **it is best practice to offer carrier screening prior to pregnancy or as early as possible in the first trimester**. Testing is available through all local pathology providers.

Some patients may be interested in more **comprehensive carrier screening**, covering hundreds (or thousands) of different genetic conditions. They may also be available through local pathology providers. Patients should be informed that out-of-pocked costs exceed $500 and results can take over 6 weeks.

If the pregnant couple are carriers for the same genetic condition, please refer **urgently** to the Department of Maternal Fetal Medicine for genetic counselling and/or invasive testing, by using the MFM referral phone 0437 537 448, or 9382 6098. Please fax all information once a referral has been made to Fax 9382 6038.

Case Example of timeframe of reproductive carrier screening in pregnancy



Patient Information: www.genetics.edu.au/ **→Pregnancy**

Where can I get more information and how do I refer?

Hospital clinical business rules: Genetic Counselling (nsw.gov.au)

Referral to the Department of Maternal Fetal Medicine - Fetal Indicaitons (nsw.gov.au)

Call the department of maternal fetal medicine and ask to speak to a genetic counsellor