

To all GPs,

CF carrier testing can be arranged by you. Therefore, Genetic Health Service - NZ is now declining referrals.

To arrange testing we would recommend:

1. You should send an EDTA blood sample to Canterbury Health Laboratories for CF panel testing, this is generally only offered to individuals with a family history of CF. This test identifies almost 90% of carriers in the Caucasian population.
2. If your patient is confirmed as a CF carrier then their partner should also have CF carrier testing, which can be arranged through their own GP.
3. If either partner is not from a Caucasian background further testing (sequencing) may be appropriate. The lab can advise you further about this if necessary.
4. If there is a known family mutation you need to provide the lab with details of this mutation (and/or the name & date of birth of the affected/carrier family member so the lab can ensure they are testing for the correct mutation).

Outcome advice:

1. If both partners are found to be CF carriers then there is a risk of them having an affected child and they should be referred to us for a discussion about this and their options in a future pregnancy.

If one partner is not a carrier of one of the mutations tested for on the panel their residual risk of being a carrier for a rare mutation is approximately 1 in 200 and the chance of them having a child with CF would then be very low.