Carrier testing for cystic fibrosis

Dear GP,

This letter contains information to support access to carrier testing for cystic fibrosis (CF). The information is taken from the **National Genomic Test Directory for rare and inherited disease** (www.england.nhs.uk/publication/national-genomic-test-directories/), which provides guidance on the eligibility criteria for different genetic tests.

A patient may be eligible for CF carrier testing (R185 in the directory) if they meet one or more of the following:

1. A family history of CF in a close relative (up to 4th degree, *ie* in 1st cousin's child or closer), or in a similar close relative of their partner
2. The partner of a known CF carrier
3. In a close relative (consanguineous) couple (1st cousins) AND from an ethnic group with a high carrier frequency (e.g. White and Northern European)
4. A prospective egg or sperm donor.

To help understand if a patient is eligible for testing, and to support the laboratory doing the test, it can be useful to collect the following information:

- For patients with a family history of CF, details of the family history including details of the CFTR pathogenic variant carried by family members (and where tested) including their exact relationship.
- For patients whose partner is affected with or a carrier of CF, details of their partner's CFTR pathogenic variant.
- The ethnicity of the patient.

Your Regional Genetics Laboratory (RGL) should accept referrals from primary care. Please find out and follow your local pathways and processes to request testing. Contact your RGL if you are unsure where to access test request forms. If you are unsure whether your patient meets the testing criteria, or have any concerns, your local Clinical Genetics service should be able to help.

If your patient has clinical symptoms suggestive of CF or a CFTR-related disorder, please refer them to the appropriate clinical specialty.

If your patient or their partner are pregnant, please make an urgent referral to Clinical Genetics that clearly states the pregnancy in the referral.

Sample requirement:

- 5ml blood sample in an EDTA tube

This letter was written with the assistance of Fiona Beecroft, Principal Genetic Counsellor, Birmingham Women's and Children's Hospital, and Anaar Sajoo, Genetic Counsellor, North West Thames Regional Genetics Service.