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Service information: Cystic Fibrosis (CF)

Gene/Locus: *CFTR* (7q31.2) **OMIM:** #219700

Referrals: Any relevant speciality e.g. Clinical Genetics, Paediatrics, Neonatal, Fertility, Respiratory
Testing: Diagnostic testing offered in clinically affected patients, carrier testing offered to patients with a family history of CF (carrier testing in patients with a family history of CF should be referred from a Clinical Genetics service and should not be under 16), carrier testing offered to parents of a fetus with echogenic bowel, carrier/diagnostic testing offered to patients undergoing fertility treatment.

Target Reporting Times:

• Diagnostic	28 calendar days	
• Carrier	28 calendar days	
• Urgent*	3 calendar days	Prenatal testing, carrier/diagnostic testing involving pregnancy

***Please contact the laboratory if urgent testing is required e.g. prenatal diagnosis, diagnostic/carrier testing when pregnancy involved**

Test Details:

- ***CFTR* mutation analysis:** ARMS PCR for the 50 most common *CFTR* mutations found in the Caucasian population using the CF-EU2v1 kit (Elucigene).

Service Details:

These mutations account for approximately 90% of *CFTR* mutations found in Caucasian English individuals in the local population. These mutations may also account for a smaller percentage of *CFTR* mutations in other ethnic groups. These mutations may also account for a smaller percentage of CF mutations in CAVD (congenital absence of the vas deferens), bronchiectasis and pancreatitis cases.

Analysis of the intron 8 splice acceptor poly T polymorphism is performed in cases filling the testing criteria (e.g. CAVD cases, patients with the p.(Arg117His) mutation, and patients with bronchiectasis or pancreatitis with one *CFTR* mutation).

It is also possible to test patients for the p.(Tyr569Asp) (Y569D) mutation. This mutation accounts for approximately 9.6% of CF mutations found in the UK Indian Sub-continent (ISC) population. (McCormick et al Eur. J. Hum Genet. (2002) 10, 583-590. This paper defines the ISC population as those of Pakistani, Indian, Bangladeshi and Asian-other origins). This test is available on request.

Sample Requirements:

- **EDTA** blood sample (1-4 ml), labelled with patient's **full name**, **date of birth** and **NHS number** or genomic DNA (20 µl at ≥30 ng/µl).
- Samples should be accompanied by a **fully** completed referral card which should include the patient's full name, date of birth, NHS number and ethnicity, if known.
- Please also include details of the test, relevant clinical details and full details of the referring clinician and centre.
- If there is a family history of CF it is important to include information about the pedigree structure and the familial mutation(s) (if known).
- Maternal samples are required in cases of prenatal diagnosis or when testing cord blood samples to exclude maternal cell contamination.

Consent:

Please note that in submitting a sample, it is the responsibility of the clinician to ensure that consent has been taken i) for testing, ii) for storage, and iii) for the use of this sample and the information generated to be shared with the patient's relatives and their health professionals. Following testing, a sample of the patient's DNA may also be used anonymously to validate new tests and for internal quality control purposes.