Cystic Fibrosis DNA Analysis

Synonyms	
Clinical Indication	CF is caused by mutations in a single large gene on chromosome 7 that encodes the cystic fibrosis transmembrane conductance regulator (CFTR) protein, which regulates chloride channel function of epithelial cells of the sweat gland, airway, pancreas, and intestine. Clinical disease occurs when disease-causing mutations are present in both copies of the CFTR gene To date over 1000 pathogenic variants with varying frequency have been identified in this gene. The ethnic origin of the patient influences the incidence of CF in the population and the pathogenic variants most commonly identified. Indications for testing include: • Confirmation of diagnosis in individuals clinically suspected of having CF. A sweat test should be undertaken prior to molecular genetic analysis wherever possible. • Testing in individuals who may have a mild variant form of CF, e.g. congenital bilateral absence of the vas deferens (CBAVD), bronchiectasis and pancreatitis. • Carrier testing in pregnant couples with foetal echogenic bowel • Carrier testing in individuals at increased risk (above the population risk) of having an affected pregnancy, for example a family history of CF, a partner shown to be a carrier or first cousin partnerships. Accurate carrier testing in CF families ideally requires either a sample from an affected family member or information regarding the pathogenic variants carried in the family. • In accordance with UK genetic testing guidelines carrier testing is only exceptionally undertaken in minors.
Part of Profile / See Also	
Request Form	Specific DNA analysis request form - http://www.labs.gosh.nhs.uk/media/764824/joint_genetics_request_form_oct_2016.pdf
Availability / Frequency of Analysis	Requests are received from Clinical Nurse Specialists in Genetics to whom patients should be referred for assessment. Other requests may be received by GP's as part of a screen for patients undergoing infertility treatment. GOSH Cystic Fibrosis Service Pack - http://www.labs.gosh.nhs.uk/media/1382012/cf_v9.pdf
Turnaround Time	4-6 weeks
Patient Preparation	
Sample Requirements	
Specimen Type	Whole Blood
Volume	Adult: 10 ml. Children: 2 - 5 ml. Babies: at least 1 ml.

Container	 PLASTIC Purple top (EDTA) tube Or  Paediatric EDTA (Red top – Sarstedt tube)  Paediatric EDTA (Lavender top BD Microtainer tube) Mix samples thoroughly for 2 minutes to prevent clotting.
Reference Range & Units	N/A
Interferences	
Interpretation & Clinical Decision Value (if applicable)	As only 50 of the most commonly identified pathogenic variants are covered by this analysis failure to identify a pathogenic variant cannot exclude affected/carrier status, a residual risk to the individual is therefore calculated and reported wherever possible. In the North European population this system detects approximately 90% of cystic fibrosis pathogenic variants. Information regarding the ethnic origin of the patient is important for calculation of residual risk as the pathogenic variant spectrum, and hence the detection rate of the assay used, varies in different populations. **Please note:** the laboratory acts only as an intermediary for genetic testing, results are sent directly to the requesting clinician. The laboratory do not receive genetic testing results and missing/overdue results must be queried directly with North East Thames Regional Genetics Laboratory Service (0207 762 6888 / genetics.labs@gosh.nhs.uk).
References	https://ukgtn.nhs.uk/find-a-test/search-by-disorder-gene/cystic-fibrosis-1/
Test code	SASO
Lab Handling	Samples are booked in as normal using the test code SASO. The top field is resulted with the coded text @GOSH. Samples should be sent to the referral laboratory the same day as collection, if this is not possible then they are placed in the fridge at 4C overnight and sent the following morning (leave a note on the handover sheet). Sent daily by courier to Regional Genetics Centre at GOSH.

UKAS MEDICAL 8692

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