

**Cystic fibrosis (CF)**      **OMIM: 219700**  
**Gene: CFTR**      **Locus: 7q31.2**      **OMIM: 602421**

**SERVICE:**      **Level 1:**      **detection of the 50 most common CFTR mutations**  
                  **Level 2:**      **mutation and dosage analysis of the CFTR gene**

**TESTING:**      **Diagnostic:** **clinically affected patients**  
                  **Carrier:**      **relatives of clinically affected patients**  
                  **Prenatal:**      **at risk of having an affected child (known mutations)**

**REFERRALS:** **from Consultant Clinicians**  
                  **The laboratory does NOT accept referrals directly from patients**

## TARGET REPORTING TIME AND COSTS

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

<b>Level 1:</b>	<b>2 weeks</b>	<b>£175 (50 common CFTR mutations)</b>
<b>Level 2:</b>	<b>8 weeks</b>	<b>£475 (sequencing and dosage)</b>
<b>Prenatal:</b>	<b>3 days</b>	<b>£350 (including maternal contamination studies)</b>

## TECHNICAL INFORMATION

- Elucigene CF EU2 kit screening for the 50 most common CFTR mutations (contact laboratory for mutation details)
- PCR and fluorescent sequence analysis of all coding exons and splice site boundaries
- Multiplex ligation dependent probe amplification analysis of all CFTR exons

## SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 50ul DNA (concentration ~500ng/ul)
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**
- Samples should be accompanied by a FULLY completed request card (available from the laboratory)
- Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital
- **Samples and paperwork must include three unique and matching patient identifiers**

## SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible.
- Do not freeze prior or during postage.

## CONSENT

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

## CONTACT DETAILS

Genetics Laboratories, Box 143  
Level 6, Addenbrooke's Treatment Centre  
Addenbrooke's Hospital  
Cambridge CB2 0QQ  
Tel: +44 (0) 1223 348866  
Fax: +44 (0) 1223 348870  
Email: [becky.treacy@addenbrookes.nhs.uk](mailto:becky.treacy@addenbrookes.nhs.uk)

Website: [www.cuh.org.uk/genetics-labs](http://www.cuh.org.uk/genetics-labs)



**Accredited Medical Laboratory**  
**Reference No: 1275**