# GENETIC TEST REPORT

**Patient Details:**

| | |
|---|---|
| Name: | John Doe |
| Date of birth: | 18 March 1995 |
| Sex: | Male |
| NHS number: | NH00198 |
| Sample type: | Blood |

**Test ordered by:**

| | |
|---|---|
| Name: | Dr Requesta |
| Organisation: | Chester Hospital |
| Telephone: | 01223 555555 |
| Copies to: | Dr A. Nother<br>Dr X. Tra |

**Test carried out by:**

| | |
|---|---|
| Laboratory: | Gentest UK |
| Telephone: | 01223 666555 |
| Date received: | 26 February 2018 |
| Date reported: | 12 March 2018 |
| Authorised by: | A Tester |

## REASON FOR TEST

CF carrier status testing requested. Partner is a carrier of CF.

## YOUR RESULT: Carrier of cystic fibrosis

## ABOUT THE TEST

This test looked at a gene called CFTR. Everyone has two copies of this gene (one from their mother and one from their father). Alterations to this gene can cause the condition cystic fibrosis (CF).

If you have an alteration in **both** copies of your CFTR genes you will have CF.

If you have an alteration in **only one** copy of CFTR you will not have CF but will be a '**carrier**'.

Carriers are healthy but may pass on their altered gene to any children.

## WHAT THIS RESULT MEANS FOR YOU

The test found that you have an alteration in one copy of your CFTR genes, making you a carrier of CF.

If you have children with someone who is also a carrier of CF, there is a 1 in 4 (25%) chance in **each pregnancy** that the child will have CF.

If you have children with someone who has not been tested for CF, there is less than 1 in 100 (less than 1%) chance that those children will have CF (some risk remains as your partner may be a carrier but not know).

In the UK population, around 1 in 25 (4%) people are carriers of CF. Because you are a carrier of CF, your close relatives have an increased chance of also being CF carriers, so carrier testing can now be offered to your adult relatives.

## NEXT STEPS

- **You can be referred to the Clinical Genetics Service to discuss your options when planning a family. Please ask your doctor for this referral if it has not been made. Take this report with you to any appointments.**
- **If your relatives would like to be tested, they should ask their GP about CF carrier testing.**

## MORE INFORMATION AND SUPPORT

The results of a genetic test can be upsetting and difficult to take in.

If you have questions about your test result, talk to the doctor who ordered your test. Your doctor can also phone or write to the East Anglian Clinical Genetics Service on 01223 216446 to make a referral or ask for further advice to help answer your queries.

To understand more about genetic testing, visit: www.nhs.uk/conditions/genetics/services/

To understand more about cystic fibrosis, visit: www.cysticfibrosis.org.uk

# FOR **YOUR RECORDS**

## Technical Information

**Patient Details:**

| | | | |
|---|---|---|---|
| Name: | John Doe | | |
| Date of birth: | 18 March 1995 | Sample type: | Blood |
| Sex: | Male | Sample collected: | 10 March 2018 |
| NHS number: | NH00198 | Sample received: | 12 March 2018 |

**The information on this page is for health professionals. It is not essential that patients read this section.**

| Gene | Zygosity | Inheritance | HGVS Description | Location | Classification |
|---|---|---|---|---|---|
| CFTR | Heterozygous | Autosomal recessive | NM_000492.3: c.1521_1523delCTT p.(Phe508del) GRCh37(hg19) | Chr7: g.117199646_117199648del | Pathogenic |

### For Your Records

The information on this page is for health professionals. It is not necessary for patients to read this section.

### Full Interpretation

John Doe's Partner, Jane (d.o.b. 28/09/2004) is a carrier of cystic fibrosis (CF) and is heterozygous for the common c.1521_1523delCTT p.(Phe508del) cystic fibrosis gene (CFTR) variant (information received with referral).

John Doe is heterozygous for the c.1521_1523delCTT p.(Phe508del) CFTR gene variant and is therefore a carrier of CF.

The risk of John Doe and Jane having a child with CF is 1 in 4 (25%) for each pregnancy . Prenatal diagnostic testing can be offered to this couple via referral to Clinical Genetics.

Carrier testing for the c.1521_1523delCTT p.(Phe508del) CFTR gene variant may be offered to John Doe's close relatives. For CF cascade testing in the East Anglian region, GPs can send a blood sample direct to the Regional Genetics Laboratory (www.cuh.nhs.uk/clinical-genetics). The patient sample form should include the name and DOB of their relative or partner who is a carrier or has CF. Relatives of the patient who live outside of East Anglia should ask their GP how testing is carried out locally.

### Test Methodology

CF-EU2v1 Elucigene Assay (http://www.elucigene.com/product/cfeu2v1/) includes 50 of the most frequently observed European CF pathogenic variants - see our website (http://cuh.org.uk/genetics-labs-cf) for variant details. Variant nomenclature is in accordance with the Human Genome Variation Society (HGVS) guidelines according to GenBank accession number NM_000492.3 (DNA) and NP_000483.3 (protein) where +1 is the A of the ATG translation initiation codon. Very rare variants within the CFTR gene may interfere with the CFEU2v1 assay, causing false positive or false negative results. Please see the Elucigene website (https://www.elucigene.com) for further information regarding CFTR analysis.

**References:**

World Health Organization (WHO), 2004. The molecular epidemiology of cystic fibrosis. Available: http://www.who.int/genomics/publications/reports/en/