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 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 was suspected due to family history 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 may now be available to your adult relatives.

NEXT STEPS

- **If you plan to have children, CF carrier testing can be offered to your partner before any pregnancy.**
- **If your relatives would like to be tested they should ask their GP about CF carrier testing.**
- **You do not need to do anything as a result of this test, but if you have questions about it, talk to your doctor.**

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

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Full Interpretation

John Doe's brother, Brian (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).

DNA from John Doe has been tested for the familial c.1521_1523delCTT p.(Phe508del) CFTR gene variant as well as other gene variants (see below). John Doe is heterozygous for the c.1521_1523delCTT p.(Phe508del) CFTR gene variant and is therefore a carrier of CF.

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 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 CF-EU2v1 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/

McCormick et al. 2002 Eur J Hum Genet. 2002 Oct;10(10):583-90.