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: No common cystic fibrosis gene alterations detected

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

Your sample has been tested for the common alterations to the CTFR gene that cause CF. No alterations were detected, so you are not a carrier of any of the alterations we tested for.

There are many alterations to the CFTR gene that are known to cause CF but some are very rare. This test looks for almost all of the alterations seen in the UK population.

Because it is possible that the CF in your family is caused by one of the rarer alterations to the CTFR gene, we cannot rule out the possibility that you carry one of these. Given the results of this test, the chance that you carry a CFTR gene alteration is about 1 in 18 (5.5%). This is only slightly higher than for the average person in the UK (4%).

In order to be more certain you could give your doctor the name, date of birth and area of residence of a relative who has CF, or is a carrier. We can then find out which CFTR alteration they have, so we can look for that same alteration in your sample. This may require an additional test.

NEXT STEPS

- **You do not need to do anything as a result of this test but if you have questions about it, talk to your doctor.**
- **If you can provide details of a relative with CF or who is a known carrier, we could investigate further.**
- **If your relatives would like to be tested, they should ask their GP about CF testing.**

MORE INFORMATION AND SUPPORT

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	No variant	Not applicable	NM_000492.3: c.1521_1523delCTT p.(Phe508del) GRCh37(hg19)	Not applicable	Not applicable

For Your Records

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Full Interpretation

John Doe has a family history of cystic fibrosis (CF), although details of the familial CFTR gene variant is not known.

DNA from John Doe has been tested for the common c.1521_1523delCTT p.(Phe508del) CFTR gene variant as well as other CFTR variants (see below) which together account for approximately 85% of UK CFTR gene variants (WHO, 2004). No CFTR gene variants were detected by our test.

This reduces John Doe's CF carrier risk to about 1 in 18 (5.5%). If the specific familial CFTR gene variant were known, we could assess John Doe's CF carrier risk directly with a targeted test.

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/