

Genetic and genomic testing is becoming increasingly widely available as testing becomes cheaper. The NHS Long Term Plan and the Improving Outcomes through Personalised Medicine effort means that nonspecialist clinicians are increasingly having to explain the results of these tests to patients. Clinicians have trouble understanding current genetic reports, let alone explaining them to patients, so we set out to create a practical template that could be rolled out at scale.



We worked with genetics specialists, non-specialist healthcare professionals, patients and members of public to design the new genetic results report template, which gives people the information they need, in a format that makes it easy to understand and know what to do next.

We started with a template for those being tested to see whether they are genetic carriers of cystic fibrosis, as this testing is now being through GPs in many cases, and GPs are often expected to explain the results to patients.



We tested this new report against ones currently used and showed that people found it easier to understand, and clearer about 'next steps'.

We followed this work with the design of a similar template for hereditary haemochromatosis.



With funding from CRUK, we then tackled the more complex test results from BRCA testing. Some BRCA gene variants affect people's chances of developing breast, ovarian or prostate cancers. BRCA testing is becoming increasingly common.

Again, after working with clinicians and patients, we tested our reports against ones currently used and found that people found it easier to understand and act on.



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: Alesford Hospital
Telephone: 01238 555555
Copies to: Dr A. Nother
Dr X. Tra

Test carried out by:

Laboratory: Acurogen UK
Telephone: 01238 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

No common cystic fibrosis gene alterations detected

Result is not framed as positive or negative

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 CFTR 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.

The fact that the test did not detect any alterations means the chance that you carry a CFTR gene alteration is around 6 in 1000 (0.6%); there is still a very small possibility that you have one of the rare alterations. However, this is highly unlikely if there is no-one in your family who has CF.

If you have children with someone who is a carrier of CF, there is a less than 2 in 1000 (0.16%) chance in **each pregnancy** that the child will have CF.

Absolute risks, percentage & frequency, same denominator

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.

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 or phone the East Anglian Clinical Genetics Service on 01238 216446. Your doctor can also phone this number for advice and 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/ or phone 01238 373 100

Three key headings patients asked for

Social support from others with same condition very important

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Technical information on page 2, so as not to distract from key messages on page 1