

GENETIC TEST REPORT

Patient Details:

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

Test requested 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

A test was requested as you have a family history of genetic haemochromatosis.

ABOUT THE TEST

This test looked for alterations in the High Iron (HFE) gene. Certain alterations to this gene can cause genetic haemochromatosis.

YOUR RESULT:

No common alterations were found. This is NOT expected to result in iron overload.

WHAT THIS RESULT MEANS

A gene is a small section of the DNA that is passed on to you from your parents. Everyone has two copies of each gene. We tested for the most common alterations found in the HFE genes. No alterations were detected. This means it is unlikely that you will develop genetic haemochromatosis.¹

¹There are some alterations to HFE genes that are known to cause genetic haemochromatosis that we are unable to test for, as these are rare. However, you are not at a substantially higher risk of having these rare alterations than the average person.

NEXT STEPS

- If you feel you need to discuss this further, or have any further questions, please speak with the person who requested your test.
- If you experience symptoms of iron overload, see your GP.

MORE INFORMATION AND SUPPORT

More information about genetic haemochromatosis can be found at:

- Haemochromatosis UK (haemochromatosis.org.uk)
- The NHS website (nhs.uk/conditions/haemochromatosis/)

FOR YOUR RECORDS

Technical Information

Patient Details:

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

This page provides technical details from the laboratory about the test.



TEST DETAILS FOR REQUESTING CLINICIAN

The information on this page provides technical details about the test.
Please keep a copy of this report your records.

RESULT SUMMARY: No variant detected

Unlikely to be affected with HFE-related GH

This patient's DNA has been tested for the common p(Cys282Tyr) (also known as C282Y) and p(His63Asp) (also known as H63D) gene alterations using allele-specific PCR.

No variant was detected.

- This patient is unlikely to be affected with HFE-related GH.
- Testing of family members is not indicated.

For more detailed guidance for health care professionals please see BSG guidelines:

bsg.org.uk/clinical-resource/diagnosis-and-therapy-of-genetic-haemochromatosis-review-and-2017-update/
or contact Clinical Genetics or the laboratory who carried out the test.

TECHNICAL DETAILS

Diplex fluorescent allele-specific PCR was used.

Nomenclature according to GenBank Accession number NM_000410.3 where +1 is the A of the ATG translation initiation codon.