

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:

You have an alteration to one of your two copies of the HFE gene, but not both. This is NOT expected to result in iron overload.

WHAT THIS RESULT MEANS

Genetic haemochromatosis, also known as hereditary haemochromatosis, is an inherited condition where iron levels in the body slowly build up over many years. This build-up of iron, known as iron overload, can cause symptoms such as feeling very tired all the time, weight loss and feeling weak. If it is not treated, this can damage parts of the body such as the liver, joints, pancreas and heart.

FOR YOU

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 have detected an alteration to one of your two copies of the HFE gene, but not both.¹ This means you are unlikely to develop genetic haemochromatosis.²

FOR YOUR FAMILY

If you have children, each child has a 50% chance of having the same alteration that you have. Your doctor can speak with you if you are planning on starting a family.

¹You only inherited this alteration from one parent, so only one of your HFE genes has it. A person with an alteration to one of their two copies of a gene is said to be "heterozygous" for that alteration.

²There 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 like those described above, see your GP.
- Family testing is not needed, but if your relatives or partner would like advice about their own risk of genetic haemochromatosis, they should speak with their GPs.

MORE INFORMATION AND SUPPORT

More information about genetic haemochromatosis and visual aids showing how it is passed on 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: Heterozygous for C282Y pathogenic variant only **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.

This patient is heterozygous for the p.(Cys282Tyr) pathogenic variant only. The p.(His63Asp) disease associated polymorphism was not detected.

- This patient is unlikely to be affected with HFE-related GH.
- Assuming that this patient has no clinical symptoms of iron overload, then this patient's risk of developing HFE-related GH is not significantly increased. In the absence of symptoms, regular biochemical monitoring of iron status is not indicated.
- Testing of family members is generally not indicated, but Clinical Genetics can be consulted if more guidance is needed.

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/](https://www.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.