GENETIC TEST REPORT

Patient Details:		Test requested by:		Test carried out by:	
Name:	Carl Doe	Name:	Dr Requesta	Laboratory:	Gentest UK
Date of birth:	18 March 1995	Organisation:	Chester Hospital	Telephone:	01223 666555
Sex:	Male	Telephone:	01223 555555	Date received:	26 February 2018
NHS number:	NH00198	Copies to:	Dr A. Nother Dr X. Tra	Date reported:	12 March 2018
Sample type:	Blood			Authorised by:	A Tester

REASON FOR TEST

A test was requested to determine if your symptoms are due to 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. Your symptoms are not likely to have been caused by this alteration.

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. 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 alteration is not likely to be the cause of your symptoms.

There 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, if a diagnosis of genetic haemochromatosis is still suspected, your doctor will refer you to a specialist.

FOR YOUR FAMILY

If you have children, each child has a 50% chance of having the same alteration that you have. Note that the particular alteration you have is not expected to result in iron overload, even if a child inherits it from both parents. Your doctor can speak with you if you are planning on starting a family.

[1]You 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.

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're still experiencing symptoms, 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		
Date of birth:	18 March 1995	Sample type:	Blood
Sex:	Male	Sample collected:	10 March 2018
NHS number:	NH00198	Sample received:	12 March 2018

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: C282Y pathogenic variant NOT 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.

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

- **We are unable to confirm a diagnosis of HFE-related GH.**
- **Not expected to cause iron overload.**
- **If a diagnosis of GH is still suspected, (i.e. raised fasting transferrin-iron saturation analysis on two or more occasions in the absence of other known causes of elevated transferrin-iron saturation) then we recommend that this patient is referred to a Consultant Hepatologist for further evaluation.**
- **Testing of family members is generally not indicated, but Clinical Genetics can be consulted if more guidance is needed.**

It is possible, but rare, that someone with a negative test does have HFE-GH. Specifically, 10% of HFE-related GH patients would not have variants detected by this test. (Note that this does not mean that 10% of people with a negative test have HFE-GH.) Please refer to BSG guidelines as below, or speak to your local testing laboratory or Clinical Genetics for more information.

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.