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:

No common alterations were found. We have not found a genetic cause for your symptoms.

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 highly unlikely that genetic haemochromatosis is causing 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.

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.**

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		
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: 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.

- **We are unable to confirm a diagnosis of HFE-related GH.**
- **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 not indicated.**

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.