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 both of your copies of the HFE gene. As you also have iron overload, this confirms a diagnosis of genetic haemochromatosis.

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 both of your copies of the HFE gene.[1] The alterations to your HFE genes mean it is likely that your symptoms are due to genetic haemochromatosis.

FOR YOUR FAMILY

As you have an alteration on both of your copies of the HFE gene, you will pass one alteration on to any children you have. Depending on the HFE genes of your partner, each child may be predisposed to genetic haemochromatosis, or they may have an alteration to one copy of the HFE gene but not both. Individuals with only one alteration are unlikely to develop genetic haemochromatosis but can pass the alteration on to their children. Your doctor can speak with you if you are planning on starting a family.

Your parents will also have alterations to one or both of their copies of the HFE gene.

Any siblings you have may also have alterations to one or both of their copies of the HFE gene.

[1] You inherited the same alteration from each parent, so both of your HFE genes have it. A person with the same alteration to both copies of a gene is said to be 'homozygous' for that alteration.

NEXT STEPS

- **If this is the first time you've learned these results, please bring these results to your GP. Please speak with your doctor about your treatment options and ask if a referral to secondary care is needed.**
- **Family testing may be available for family members and your partner. Individuals wanting testing should speak with their GPs.**

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

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WHAT CAN BE DONE TO MANAGE GENETIC HAEMOCHROMATOSIS?

There's currently no cure for genetic haemochromatosis, but there are treatments that can reduce the amount of iron in your body. This can help relieve some of the symptoms and reduce the risk of damage to organs such as the heart, liver and pancreas. We know that if genetic haemochromatosis is diagnosed early and treatment started, serious complications can be avoided and patients can have a normal life expectancy.

TREATMENT

The most commonly used treatment for genetic haemochromatosis is a procedure to remove some of your blood, known as a phlebotomy or venesection. The removed blood includes red blood cells that contain iron, and your body will use up more iron to replace them, helping to reduce the amount of iron in your body. If it's difficult for you to have blood removed regularly, for example if you have very thin or fragile veins, then your doctor may discuss a different treatment called chelation therapy.

LIFESTYLE

You do not need to make any big changes to your diet, such as avoiding all foods containing iron, if you have genetic haemochromatosis. This is unlikely to be of much extra help if you're having one of the treatments above and could mean you do not get all the nutrition you need.

However, there are some dietary changes you can make regarding avoiding certain foods or supplements, and limiting alcohol consumption.

Reducing iron in your diet may slow the continued iron build-up but cannot remove iron that has already accumulated nor replace proper treatment.

Maintaining a healthy weight, being physically active and not smoking are also important for staying healthy.

More details can be found here: https://www.nhs.uk/conditions/haemochromatosis/treatment/.

MORE INFORMATION AND SUPPORT

More information and support for individuals facing genetic haemochromatosis and visual aids showing how it is passed on can be found at:

- Haemochromatosis UK (haemochromatosis.org.uk) - Helpline: 03030 401 102 (weekdays 12pm-3pm)
- The NHS website (nhs.uk/conditions/haemochromatosis/)

You can also find more information about genetic haemochromatosis and other problems it can cause on the British Heart Foundation, Diabetes UK and British Liver Trust websites.

FOR **YOUR RECORDS**

Technical Information

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

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

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: Homozygous for C282Y pathogenic variant
Confirms a diagnosis of HFE-related GH in the presence of iron overload

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 homozygous for the p.(Cys282Tyr) pathogenic variant.

- **This result is consistent with the presence of iron overload and confirms a diagnosis of HFE-related GH.**
- **We recommend that indices of iron overload (serum transferrin saturation and ferritin) are regularly monitored and managed as per our local guidelines:**
cuh.nhs.uk/clinical-genetics/service-information/general-guidelines-for-health-professionals/haemochromatosis
- **Symptomatic patients may also be investigated according to their symptoms.**
- **Testing is recommended for siblings and adult children, if they have not already been tested and if they are not already known carriers. Carrier testing for parents and testing of the partner to establish risk to children is also recommended. Carrier testing for C282Y may be available for extended family members.** For more information, speak to your local testing laboratory or Clinical Genetics.

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.