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 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 both of your copies of the HFE gene. This indicates you are predisposed to 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] This means you are predisposed to genetic haemochromatosis and are at risk of developing iron overload. In one study[2], over 50% of people with these alterations showed signs of iron overload. This compared with about 1% of people without these alterations. For this reason, your doctor may suggest that your iron levels be monitored, and may discuss treatment if your iron levels are high. With treatment, most people will not suffer ill effects from high iron levels.

FOR YOUR FAMILY

As you have an alteration to 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 also 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.
[2] Allen, Katrina J., et al. "Iron-overload–related disease in HFE hereditary hemochromatosis." NEJM 358.3 (2008): 221-230. In this study, over 50% of C282Y homozygotes showed at least provisional evidence of iron overload.

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 what this result means for you and ask if a referral to secondary care is needed. Your GP may suggest that you have a blood test to find out whether further action is recommended.**
- **If you experience symptoms like those described above, see your GP.**
- **Family testing may be available for family members and your partner. Individuals wanting testing should speak with their GPs.**

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WHAT CAN BE DONE ABOUT MY RISK?

Your doctor may recommend a blood test to check the amount of iron in your body. Depending on the results, they may recommend that your iron levels be monitored regularly, or may suggest treatments that can reduce the amount of iron. This can reduce the risk of serious complications which can affect organs such as the heart, liver and pancreas.

The important thing is to be aware of the symptoms of genetic haemochromatosis. You may never develop any symptoms at all. If symptoms do occur, they usually begin between the ages of 30 and 60, although they can also happen earlier.

The symptoms tend to develop earlier in men than in women. Women often do not experience problems until after the menopause.

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.

Initial symptoms of genetic haemochromatosis can include:

- feeling very tired all the time (fatigue)
- weight loss
- weakness
- joint pain
- an inability to get or maintain an erection (erectile dysfunction)
- irregular periods or absent periods

These symptoms can have many different causes but if you have persistent or worrying symptoms, please see your GP.

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. 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 can be 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
At increased risk of developing symptoms of 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 homozygous for the common p.(Cys282Tyr) pathogenic variant.

- **This patient is at increased risk of developing symptoms of HFE-related GH. In women of reproductive age these rarely present before menopause.**
- **We recommend routine screening of 09:00am fasting transferrin saturation and ferritin and referral for treatment as appropriate. More information can be found under "Family Screening" of our local guidelines:** cuh.nhs.uk/clinical-genetics/service-information/general-guidelines-for-health-professionals/haemochromatosis
- **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.