

Report to	IMEDICAL, 1 Union st , Pyrmont, NSW, 2009	Patient	BROWN, AMANDA C/O IMEDICAL 1 UNION ST PYRMONT NSW 2009	Phone	0427594760	Age	57 years	Sex	F
		D.O.B	08/11/1966	Collect date	05/12/2023	Lab ref	23-25594757		
				Collect time	08:27 AM	Your ref			
				Reported	14/12/2023		10:43 AM		
Tests requested	HGA, SCP, TFT, DVI, MBA, LIP, FE, GLU, VBF, FBE								

Clinical notes

HEREDITARY HAEMOCHROMATOSIS GENOTYPING

Specimen:
Blood

Result:
C282Y HFE Gene Mutation: Not Detected

H63D HFE Gene Mutation: **DETECTED HETEROZYGOUS**

S65C HFE Gene Mutation: Not Detected

Comments:
This sample is heterozygous for the H63D mutation which is generally associated with a normal phenotype.

Hereditary Haemochromatosis (HH) is a predominantly autosomal recessive disorder of the HFE gene. Greater than 90% of HH patients have a homozygous expression of a single mutation (C282Y) in this gene. Those who are heterozygous for C282Y are carriers but very rarely develop significant iron overload.

Other mutations in the HFE gene have also been described, the most common of which is H63D. Compound heterozygotes with one copy of H63D and one copy of C282Y may develop iron overload. This is usually not as severe as in C282Y homozygotes but may be clinically significant. Homozygotes for H63D or S65C may have iron overload in some cases, usually mild. Heterozygotes are very unlikely to have iron overload.

Treatment decisions should always be based on clinical features and iron studies as well as genetic test results, especially in cases involving mutations other than the homozygous expression of C282Y.

Family studies should be performed if a patient is homozygous for C282Y. With other genotypes family studies are less likely to detect individuals at high risk.

Dr Kym Mina MBBS PhD FRCPA

Clinical Director, Genomic Diagnostics

SURGERY USE

Normal

No Action/File

Patient
Notified

Make
Appoint.

Further Tests

Notes
Required

Speak
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SERUM HIGH SENSITIVITY C-REACTIVE PROTEIN (CRP)

Request Number	25594757
Date Collected	5 Dec 23
Time Collected	08:27
hsCRP (< 4.91) mg/L	2.31

The CDC / AHA recommend the following hsCRP cut-off points (tertiles) for cardiovascular disease risk assessment:

hsCRP level (mg/L)	Relative Risk
<1.0	Low
1.0 - 3.0	Average
>3.0	High

The average of two CRP tests, ideally taken two weeks apart, produces a more stable estimate of this marker.

A CRP greater than 10mg/L should prompt a search for a source of infection or inflammation.

THYROID PROFILE

Specimen Type: Serum			
TSH	1.4	mIU/L	(0.5-4.0)
FT4	16	pmol/L	(10-20)
FT3	5.6	pmol/L	(3.5-6.5)

Result(s) consistent with euthyroidism.

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VITAMIN D

Haemolysis	Nil
Serum 25(OH) Vitamin D	68 nmol/L

Suggested decision limits for Vitamin D status:

Sufficiency	51 -200	nmol/L
Mild deficiency	25 - 50	nmol/L
Marked deficiency	< 25	nmol/L
Toxicity	>250	nmol/L

References: Vitamin D and health in adults in Australia and New Zealand:
Position Statement. MJA 2012 June 18; 196(11),686-687.

SERUM CHEMISTRY

Specimen Type: Serum

Haemolysis	Nil		
Icterus	Nil		
Lipaemia	Nil		
Sodium	144	mmol/L	(135-145)
Potassium	4.6	mmol/L	(3.6-5.4)
Chloride	103	mmol/L	(95-110)
Bicarbonate	24	mmol/L	(22-32)
Anion Gap	22	mmol/L	(10-20)
Urea	7.3	mmol/L	(2.5-9.0)
Creatinine	70	umol/L	(45-90)
eGFR	80		mL/min/1.73m^2
Urate	0.37	mmol/L	(0.14-0.36)
Bilirubin	9	umol/L	(< 15)
AST	29	U/L	(< 35)
ALT	27	U/L	(< 30)
GGT	33	U/L	(< 35)
Alkaline Phosphatase	75	U/L	(30-115)
Protein	70	g/L	(60-82)
Albumin	45	g/L	(38-50)
Globulin	25	g/L	(20-39)
Calcium	2.45	mmol/L	(2.10-2.60)
Corrected Calcium	2.41	mmol/L	(2.10-2.60)

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Phosphate	1.57	mmol/L	(0.75-1.50)
Creatine Kinase	56	U/L	(< 211)
Magnesium	0.81	mmol/L	(0.70-1.10)

eGFR values between 60 and 89 mL/min/1.73m² should be interpreted with caution. These results are only consistent with CKD in the presence of other evidence such as microalbuminuria, proteinuria or haematuria.
Ref:Lamb EJ etal in Ann Clin Biochem 2005; 42:321-345.

LIPID STUDIES

Specimen Type: Serum

Reference intervals are included for reference only, and interpretation / treatment goals should be guided by patient-specific cardiovascular risk assessment (see Australian Cardiovascular Risk Charts. Alternatively, the web-site www.cvdcheck.org.au can be accessed in order to complete a risk assessment for individual patients.)

Haemolysis	Nil
Icterus	Nil
Lipaemia	Nil

Fasting status	Fasting
Total Cholesterol	6.0 mmol/L (3.9-5.2)
Triglycerides	0.6 mmol/L (0.5-1.7)
HDL Cholesterol	2.0 mmol/L (1.0-2.0)
LDL Cholesterol	3.7 mmol/L (1.5-3.4)
Non-HDL Cholesterol	4.0 mmol/L (< 3.4)
Cholesterol/HDL-C Ratio	3.0 (< 4.5)

NVDPa TARGET LIPID RANGES (MMOL/L) FOR PATIENTS AT HIGH / MODERATE RISK OF CARDIOVASCULAR DISEASE:

TOTAL CHOLESTEROL	<4.0
TRIGS (FASTING)	<2.0
HDL-C	>= 1.0
LDL-C	<2.0

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| NON HDL-C | <2.5 |

LDL-C exceeds target for higher risk patients and may be excessive in some individuals.

IRON STUDIES

Specimen Type: Serum			
Serum Iron	18	umol/L	(10-30)
Transferrin	26	umol/L	(32-48)
Transferrin Saturation	35	%	(13-45)
Serum Ferritin	231	ug/L	(30-400)

Transferrin may be decreased by inflammation (acute or chronic), or protein deficiency or loss. The ferritin concentration excludes iron deficiency.

SERUM/PLASMA GLUCOSE

Fasting status	Fasting		
Serum	5.0	mmol/L	(3.4-5.4)

Normal glucose concentration.

VITAMIN B12 AND FOLATE STUDIES

Vitamin B12	386	pmol/L	(156-740)
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, Pyrmont, NSW, 2009

Patient **BROWN, AMANDA**
C/O IMEDICAL 1 UNION ST PYRMONT NSW
2009

Phone 0427594760
D.O.B 08/11/1966 Age 57 years Sex F

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HAEMATOLOGY

Date Collected 05 Dec 23
Time Collected 08:27
Specimen Type: EDTA

Hb	141	g/L	(115-165)	WBC	6.5	x10 ⁹	/L (4.0-11.0)
RCC	4.8	x10 ¹²	/L (3.9-5.8)	Neut	3.4	x10 ⁹	/L (2.0-7.5)
Hct	0.43		(0.34-0.47)	Lymp	2.1	x10 ⁹	/L (1.0-4.0)
MCV	90	fL	(79-99)	Mono	0.6	x10 ⁹	/L (0.2-1.0)
MCH	30	pg	(27-34)	Eos	0.3	x10 ⁹	/L (< 0.7)
MCHC	329	g/L	(320-360)	Baso	0.1	x10 ⁹	/L (< 0.2)
RDW	12.1	%	(10.0-17.0)				
Plat	285	x10 ⁹	/L (150-400)				

HAEMATOLOGY: FBC parameters are within reference range.

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