

PYATT, ANGELA
21 BROOKS ROAD, MAREEBA. 4880
Phone: 0492469270
Birthdate: 06/09/1991 Sex: F Medicare Number: 4336253244
Your Reference: 427A24D8F1 Lab Reference: 527747803-D-D010
Laboratory: SNP
Addressee: DR HEIKE KATH Referred by: DR HEIKE KATH
Name of Test: HFE
Requested: 18/07/2024 Collected: 23/07/2024 Reported: 30/07/2024
12:51

Clinical notes: high transferrin saturation

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Haemochromatosis Genotyping

Specimen	EDTA Blood
C282Y	* Heterozygous Mutation Detected
H63D	Mutation Not Detected

Please note these variants are known as c.845G>A, p.Cys282Tyr, c.187C>G, p.His63Asp, and when reported c.193A>T, p.Ser65Cys, according to HGVS nomenclature; RefSeqGene: NG_008720.1 (HFE).

Comments on Collection 527747803

This patient is a heterozygous carrier of the HFE: c.845G>A, p.Cys282Tyr mutation. This genotype is NOT consistent with a diagnosis of HFE-associated haemochromatosis.

Note that elevated ferritin can be seen in inflammatory states (e.g. infection, autoimmune disease or malignancy), or as a result of hepatic pathology (e.g. viral hepatitis, alcohol-related liver disease and non-alcoholic fatty liver disease).

If there is clinical suspicion of haemochromatosis (persistent hyperferritinaemia and transferrin saturation>45%), referral to a Gastroenterologist or Haematologist may be indicated, as there are other rare causes of iron overload and haemochromatosis.

This result may have implications for other family members. Testing for at-risk relatives is available, and genetic counselling may be useful.

Test Information:

Testing was performed utilising endpoint genotyping on the LC480 and pyrosequencing of codons 63 to 65 of the HFE gene, where required. This result does not exclude mutations being present at other locations within the HFE gene (RefSeq: NG_008720.1).

NPAAC guidelines suggest that repeat testing on a separately collected specimen may be warranted for genetic tests to identify any potential errors in specimen collection, testing or reporting. Repeat testing is especially recommended if the results will impact significantly on clinical management, or if the results are inconsistent with family history, clinical features or other laboratory results.

For clinician enquiries regarding these results, please contact Dr James Harraway (07 3377 8666).

AC

Sullivan Nicolaides Pty Ltd. ABN 38 078 202 196. NATA/RCPA Accreditation No
1964

Tests Completed: HFE PCR
Tests Pending :
Sample Pending :

PYATT, ANGELA
21 BROOKS ROAD, MAREEBA. 4880
Phone: 0492469270
Birthdate: 06/09/1991 Sex: F Medicare Number: 4336253244
Your Reference: 4365618CF2 Lab Reference: 529186898-C-H245
Laboratory: SNP
Addressee: DR JAIMELEE B MILLINER Referred by: DR JAIMELEE B MILLINER

Name of Test: .ANAEMIA
Requested: 06/08/2024 Collected: 06/08/2024 Reported: 07/08/2024
12:52

Haematinics

Date	Latest Results			Reference	Units
	27-Mar-19	28-Jun-24	06-Aug-24		
Time	1041	1056	1446		
Lab Id.	647274136	686794467	529186898		
Iron	26	31 H	24	(5-30)	umol/L
Transferrin	2.3	2.3	2.2	(1.9-3.1)	g/L
TIBC	58	57	56	(47-77)	umol/L
Trans Sat	45	54 H	43	(20-45)	%
Ferritin	30	44	43	(30-250)	ug/L
CRP		0.7		(<5)	mg/L
Vitamin B12		286		(>150)	pmol/L
Active B12		100		(>35)	pmol/L
Folate serum		39		(>7.0)	nmol/L

Comments on Collection 06-Aug-24 1446:
Iron Studies
Normal Iron Status.

CA

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Tests Completed: Iron Studies
Tests Pending :
Sample Pending :

