RCPA PITUS

RCPA NATA WORLD RECOGNISED ACCREDITATION

PITUS Pathology

NATA Accreditation # 9999

MANNING, Norman Lyle (Mr)	Sex **Male**	Request ID **1978881777**
84 Nakiska Court	DOB **23-May-68, aged 52y**	on **20-Sep-20**
Sunrise Bay QLD 4573		by **Dr Trish FAMILYDR**
	MRecord **719909917**	733 Little Archie Street
	Located at Sunrise Hospital	Brisbane QLD 4001
	Metabolic Clinic	Provider 316422R

Haemochromatosis gene screening – Final Report

Clinical history – ? Hereditary haemochromatosis. Arthritis, increased iron stores

Report issued 20-Sep-20 08:20

	Latest results	Reference	Units
Collection date	**20-Sep-20**		
Collection time	**08:20**		
Request ID	**1978881777**		
Specimen	**Blood**		
C28Y mutation analysis	Not Detected		
H63D mutation analysis	Homozygous		

Results

Two copies of the p.His63Asp variant were detected in the patient. The p.Cys282Tyr variant was not detected. The diagnosis of the most common form of HFE-related hereditary haemochromatosis is excluded.

Interpretation

Hereditary haemochromatosis (HH) is a recessive genetic disorder of iron metabolism. Greater than 90% of HFE related hereditary haemochromatosis is associated with homozygosity for p.(Cys282Tyr) (c.845G>A; aka p.C282Y). Compound heterozygosity for p. (Cys282Tyr) and p. (His63Asp) (c.187C>G; aka p.H63D) may be a risk factor predisposing to mild to moderate forms of iron overload when in association with other risk factors. Both mutations are detected by real-time PCR amplification and fluorescent detection of alleles.

Dr Kondos Pathologist
+61 7 3662 9871

Copy to 1 - Genetics Clinic
Copy to 2 - Dr Bjorn Genetic Counsellor, 81 Estelle Parade, New Farm
Copy to 3 - MyHealthRecord, IHI 8003 6088 3335 7361

For consumer information on pathology results visit Lab Tests Online Australia https://www.labtestsonline.org.au/