

PATIENT INFORMATION		REFERRAL INFORMATION		
NAME JANE DOE		CLINIC NAME CLINIC X		
ETHNICITY ETHNICITY X		CLINIC ID XXXX		
DATE OF BIRTH XX/XX/XXXX		REFERRING CLINICIAN Dr. XXXX		
TEST INDICATIONS XXXXXXXX		CLINIC EMAIL XXXXXX@email.com		
SAMPLE INFORMATION				
ORDER NUMBER XXXX	LAB NUMBER XXXX	DATE OF COLLECTION XX/XX/XXXX	DATE RECEIVED XX/XX/XXXX	
RODINIA INFERTILITY TEST				
PANEL SELECTED*		FEMALE INFERTILITY PANEL <input checked="" type="checkbox"/> MALE INFERTILITY PANEL <input type="checkbox"/>		
ADD-ON PANEL SELECTED		YES <input type="checkbox"/> NO <input checked="" type="checkbox"/>		
TEST RESULTS				
Clinically Significant Findings / Variant Detected				
NUMERICAL / STRUCTURAL ABNORMALITIES				
RESULTS	No numerical or structural abnormalities detected			
ZYGOSITY	VARIANT DETECTED	AMINO ACID CHANGE	GENE	INHERITANCE
Heterozygous	ENST00000397262.1:1 c.17G>A	p.(Arg6His)	INS	Autosomal Dominant
INTERPRETATION				
<p>Variant Summary: A heterozygous c.17G>A variant was detected at exon 1 of the INS gene (Ensembl ID: ENST00000397262.1). This is a missense substitution as it changes the amino acid sequence to include at position 6 of the protein sequence. The amino acid change is conservative in terms of the physicochemical properties of these amino acids. This variant is within a functional domain of the protein with no change variation. It is a missense variant in a gene that has a low rate of benign variation and in which missense variants are a common mechanism of the disease. In addition, this variant is found in population databases in frequency less than what is expected for a dominant allele. An alternative variant has been described, Arg7Cys and has been described as pathogenic by Segre¹ and confirmed using WES. It was in silico predicted to be pathogenic using prediction for this variant versus one pathogenic prediction. Based on the information described above and according to ACMG guidelines this variant is classified as likely pathogenic.</p> <p>Clinical Significance: The INS gene provides instructions for producing the hormone insulin, which is necessary for the normal regulation of glucose levels in the blood. Insulin is a single chain and the primary energy source</p>				

Polycystic ovary syndrome (PCOS) is the most common endocrine disorder in women of reproductive age. It is characterized by irregular menstrual cycles, excessive hair growth, and acne. PCOS is a complex condition and genetic testing can help identify individuals who are at a higher risk of developing the syndrome. Early diagnosis and treatment can help manage the symptoms and reduce the risk of long-term complications such as type 2 diabetes and heart disease.

Recommendation: This individual is heterozygous carrier for the c.2704G variant in the FTO gene. Genetic testing is recommended for all individuals undergoing genetic testing. Clinical correlation with other clinical findings is advised.

ADD-ON PANEL: HAEMOPHILIA and THROMBOSIS

NOT SELECTED

METHODOLOGY/LIMITATIONS

Rodinia is a Laboratory Developed Test (LDT) that uses Next-Generation Sequencing (NGS) for identifying variants. Genetic testing is performed using a high-throughput sequencing platform that allows for the simultaneous analysis of multiple genes. The test is performed using a library of DNA fragments that are sequenced and analyzed using a bioinformatics pipeline. The test is designed to identify variants in the FTO gene and other genes associated with PCOS. The test is performed using a high-throughput sequencing platform that allows for the simultaneous analysis of multiple genes. The test is performed using a library of DNA fragments that are sequenced and analyzed using a bioinformatics pipeline. The test is designed to identify variants in the FTO gene and other genes associated with PCOS.

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Genetic testing is performed using a high-throughput sequencing platform that allows for the simultaneous analysis of multiple genes. The test is performed using a library of DNA fragments that are sequenced and analyzed using a bioinformatics pipeline. The test is designed to identify variants in the FTO gene and other genes associated with PCOS. The test is performed using a high-throughput sequencing platform that allows for the simultaneous analysis of multiple genes. The test is performed using a library of DNA fragments that are sequenced and analyzed using a bioinformatics pipeline. The test is designed to identify variants in the FTO gene and other genes associated with PCOS.

... (faint text) ...

The Rodinia infertile test development and performance evaluation was carried out by NIPD Genetics in its own laboratory, which is registered under the Clinical Laboratory Agreement Act of 2008 (2008) in Cyprus. The laboratory is currently testing Rodinia's infertile test and other genetic tests registered as infertile tests in Cyprus. The test results will be reported to you by NIPD Genetics and they will be available to you through your doctor's office.

ADDITIONAL TECHNICAL SPECIFICATIONS

... (faint text) ...

ADDITIONAL INFORMATION / DISCLOSURE

... (faint text) ...

SUPPLEMENTARY INFORMATION

Disease (Gene)	Results
Fragile X (FMR1)	Allele1: 30 repeats Allele2: 33 repeats

REFERENCES

1. Fennell, T. J. et al. Variation across 282,952 human exomes and genomes reveals the spectrum of loss-of-function intolerance across human protein-coding genes. *bioRxiv* (2015) doi:10.1101/012121
2. Saitou, N. et al. InfPath: The universal protein knowledgebase. *Nucleic Acids Res* (2017) doi:10.1093/nar/gkx1215

1. Richards, S. et al. Standards and guidelines for the interpretation of sequence variants: A joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med* (2015) doi:10.1038/gim.2015.24.
2. US Department of Health and Human Services. *Genetics Home Reference*. U.S. National Library of Medicine (2015).
3. Harada, A., Scott, A. F., Anderson, J., Valle, D., & McPherson, V. A. Online Mendelian Inheritance in Man (OMIM). *Hum Mutat* (2002) doi:10.1002/9781118130461.ch107.40-4444.121-12.10.2.4.

Approved by:

Approved by:

Date of report (DD/MM/YYYY):

REPORT EXAMPLE

Table 1 - Genes tested by the Rodinia Infertility Panel

FEMALE INFERTILITY PANEL				
AIRE	EIF2B3	GALT	IRS2	PROKR2
ANOS1	FEZF1	GDF9	KISS1	PSMC3IP
BMP15	FGF17	GNAS	KISS1R	SEMA3A
CAPN10	FGF8	GNRH1	LHB	SPRY4
CHD7	FGFR1	GNRHR	LHCGR	STAG3
CYP11A1	FIGLA	HESX1	NOBOX	TAC3
CYP17A1	FLRT3	HS6ST1	NR5A1	TACR3
CYP19A1	FMR1	IL17RD	NSMF	THADA
DENND1A	FOXL2	INS	POF1B	WDR11
DUSP6	FSHB	INSR	POLG	WT1
EIF2B2	FSHR	IRS1	PROK2	ZP1

MALE INFERTILITY PANEL				
ANOS1	DUSP6	FSHR	LHB	SPRY4
AR	FEZF1	GNRH1	LHCGR	SRD5A1
AURKC	FGF17	GNRHR	NR5A1	SRY
CATSPER1	FGF8	HESX1	NSMF	TAC3
CFTR	FGFR1	HS6ST1	PRM1	TACR3
CHD7	FLRT3	IL17RD	PROK2	USP26
DAZL	FMR1	KISS1	PROKR2	USP9Y
DDX25	FSHB	KISS1R	SEMA3A	WDR11

REPORT EXAMPLE

THROMBOPHILIA AND NAIT PANEL

Disorder / Common name	Gene	Variant	Alternative nomenclature
Factor V Leiden	F5	NM_000130.4(F5):c.1601G>A (p.Arg534Gln)	G1691A F5,ARG506GLN R506Q Factor V Leiden
Factor V R2	F5	NM_000130.4(F5):c.3980A>G (p.His1327Arg)	FV R2 H1299R A4070G R2 allele
Factor XIII	F13A1	NM_000129.3(F13A1):c.103G>T (p.Val35Leu)	p.Val34Leu F13A1; VAL34LEU; V34L
HPA-1	ITGB3	NM_000212.2(ITGB3):c.176T>C (p.Leu59Pro)	L33P
HPA-2	GP1BA	NM_000173.7(GP1BA):c.482C>T (p.Thr161Met)	rs6065
HPA-3	ITGA2B	NM_000419.5(ITGA2B):c.2621T>G (p.Ile874Ser)	I843S
HPA-4	ITGB3	NM_000212.2(ITGB3):c.506G>A (p.Arg169Gln)	R143Q
HPA-5	ITGA2	NM_002203.4(ITGA2):c.1600G>A (p.Glu534Lys)	Not available
HPA-6	ITGB3	NM_000212.2(ITGB3):c.1544G>A (p.Arg515Gln)	R489Q
PAI-1 4G/5G	SERPINE1	NM_000602.5(SERPINE1):c.-820G[(4_5)]	4G/5G
MTHFR	MTHFR	NM_005957.5(MTHFR):c.665C>T (p.Ala222Val)	C677T; MTHFR; 677C-T; ALA222VAL (rs1801133)
MTHFR	MTHFR	NM_005957.4(MTHFR):c.1286A>C (p.Glu429Ala)	MTHFR; 1298A-C; GLU429ALA (rs1801131)
ACE (I/D)	ACE	NM_000789.3(ACE):c.2306-117_2306-116insAF118569.1: g.14094_14382	ACE/ID polymorphism INS/DEL (rs1799752)
Apo B	APOB	NM_000384.3(APOB):c.10580G>A (p.Arg3527Gln)	R3500Q 9775G>A
Apo E	APOE	NM_000041.2(APOE):c.526C>T (p.Arg176Cys)	R158C R148C
Apo E	APOE	NM_000041.4(APOE):c.388T>C (p.Cys130Arg)	C112R ApoE4
MTR	MTR	NM_000254.2(MTR):c.2756A>G (p.Asp919Gly)	p.D919G:GAC>GGC 2756A-G
MTRR	MTRR	NM_002454.3(MTRR):c.66A>G (p.Ile22Met)	p.I49M:ATA>ATG
AGT	AGT	NM_000029.4(AGT):c.803T>C (p.Met268Thr)	M235T NM_000029.3:c.803T>C
AGTR1	AGTR1	NM_031850.3(AGTR1):c.*86A>C	A1166C
GSTP1	GSTP1	NM_000852.4(GSTP1):c.313A>G (p.Ile105Val)	rs1695 GSTP1*B
Prothrombin	F2	NM_000506.5(F2):c.*97G>A	F2 rs1799963 20210G-A G20210A