NIPD GENETICS
A MEDICOVER COMPANY

Rodinia®
infertility test

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	LAB NUMBER	DATE OF COLLECTION	DATE RECEIVED
XXXX	XXXX	XX/XX/XXXX	XX/XX/XXXX

RODINIA INFERTILITY TEST

PANEL SELECTED*	FEMALE INFERTILITY PANEL ☒ MALE INFERTILITY PANEL ☐
ADD-ON PANEL SELECTED	YES ☐ NO ☒

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

[redacted]

ADD-ON PANEL: HAEMOPHILIA and THROMBOSIS

NOT SELECTED

METHODOLOGY/LIMITATIONS

ADDITIONAL TECHNICAL SPECIFICATIONS

ADDITIONAL INFORMATION / DISCLOSURE

SUPPLEMENTARY INFORMATION

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

REFERENCES

3. 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.2015.30.

4. U.S Department of Health and Human Services. Genetics Home Reference. U.S Natl. Libr. Med. (2017).

5. Hamosh, A., Scott, A. F., Amberger, J., Valle, D. & McKusick, V. A. Online Mendelian Inheritance in Man (OMIM). Hum. Mutat. (2000) doi: 10.1002/(SICI)1098-1004(2000)15:1<57::AID-HUMU12>3.0.CO;2-G.

Approved by:

Approved by:

Date of report (DD/MM/YYYY):

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

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