

## RODINIA INFERTILITY PANEL

### FEMALE INFERTILITY PANEL

Testing of 55 genes and whole, partial and mosaic sex chromosome aneuploidies.

Disorders tested include primary ovarian insufficiency, ovarian hyperstimulation syndrome and hypogonadotropic hypogonadism such as Kallmann syndrome.

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

Testing of 40 genes and whole, partial and mosaic sex chromosome aneuploidies, including Y chromosome microdeletions.

Disorders tested include hypogonadotropic hypogonadism such as Kallmann syndrome.

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

Tests for 22 genetic variants in 17 genes. Can be selected as an Add-on Panel to the Female or Male Panel, or as a Stand-alone Panel.

DISORDER / COMMON NAME	GENE	VARIANT	ALTERNATIVE NOMENCLATURE
<b>Factor V Leiden</b>	F5	NM_000130.4(F5):c.1601G>A (p.Arg534Gln)	G1691A F5, ARG506GLN R506Q Factor V Leiden
<b>Factor V R2</b>	F5	NM_000130.4(F5):c.3980A>G (p.His1327Arg)	FV R2 H1299R A4070G R2 allele
<b>Factor XIII</b>	F13A1	NM_000129.3(F13A1):c.103G>T (p.Val35Leu)	p.Val34Leu F13A1 VAL34LEU V34L

DISORDER / COMMON NAME	GENE	VARIANT	ALTERNATIVE NOMENCLATURE
<b>HPA-1</b>	ITGB3	NM_000212.2(ITGB3):c.176T>C (p.Leu59Pro)	L33P
<b>HPA-2</b>	GP1BA	NM_000173.7(GP1BA):c.482C>T (p.Thr161Met)	rs6065
<b>HPA-3</b>	ITGA2B	NM_000419.5(ITGA2B):c.2621T>G (p.Ile874Ser)	I843S
<b>HPA-4</b>	ITGB3	NM_000212.2(ITGB3):c.506G>A (p.Arg169Gln)	R143Q
<b>HPA-5</b>	ITGA2	NM_002203.4(ITGA2):c.1600G>A (p.Glu534Lys)	Not available
<b>HPA-6</b>	ITGB3	NM_000212.2(ITGB3):c.1544G>A (p.Arg515Gln)	R489Q
<b>PAI-1 4G/5G</b>	SERPINE1	NM_000602.5(SERPINE1):c.-820G[[4_5]]	4G/5G
<b>MTHFR</b>	MTHFR	NM_005957.5(MTHFR):c.665C>T (p.Ala222Val)	C677T MTHFR 677C-T ALA222VAL (rs1801133)
<b>MTHFR</b>	MTHFR	NM_005957.4(MTHFR):c.1286A>C (p.Glu429Ala)	MTHFR 1298A-C A1298C GLU429ALA (rs1801131)
<b>ACE (I/D)</b>	ACE	NM_000789.3(ACE):c.2306-117_2306-116insAF118569.1: g.14094_14382	ACE/ID polymorphism INS/DEL (rs1799752)
<b>Apo B</b>	APOB	NM_000384.3(APOB):c.10580G>A (p.Arg3527Gln)	R3500Q 9775G>A
<b>Apo E</b>	APOE	NM_000041.2(APOE):c.526C>T (p.Arg176Cys)	R158C R148C
<b>Apo E</b>	APOE	NM_000041.4(APOE):c.388T>C (p.Cys130Arg)	C112R ApoE4
<b>MTR</b>	MTR	NM_000254.2(MTR):c.2756A>G (p.Asp919Gly)	p.D919G:GAC>GGC 2756A-G
<b>MTRR</b>	MTRR	NM_002454.3(MTRR):c.66A>G (p.Ile22Met)	p.I49M:ATA>ATG
<b>AGT</b>	AGT	NM_000029.4(AGT):c.803T>C (p.Met268Thr)	M235T NM_000029.3:c.803T>C
<b>AGTR1</b>	AGTR1	NM_031850.3(AGTR1):c.*86A>C	A1166C
<b>GSTP1</b>	GSTP1	NM_000852.4(GSTP1):c.313A>G (p.Ile105Val)	rs1695 GSTP1*B
<b>Prothrombin</b>	F2	NM_000506.5(F2):c.*97G>A	F2 rs1799963 20210G-A G20210A