GENE VARIANTS ASSOCIATED WITH REPRODUCTION DISORDERS V4

- LIST OF EXAMINED DISORDERS

Gene name	OMIM gene number	Target variant (HGVS nomenclature)	Comment
F2	176930	c.*97G>A	"Prothrombin" mutation; Inherited thrombophilia
F5	612309	c.1601G>A (p.Arg534Gln)	"Leiden mutation"; Inherited thrombophilia
MTHFR	607093	c.665C>T (p.Ala222Val) (alternatively described as c. 677C>T)	Risk of neural tube defects in foetus (for women); Inherited thrombophilia; Risk of miscarriages due to lower fetal viability
		c.1286A>C (p.Glu429Ala) (alternatively described as c.1298A>C)	Risk of miscarriages due to lower fetal viability
ANXA5	131230	c.-229G>A c.-210A>C c.-184T>C c.-135G>A	Analysis of M2/M1 haplotypes; Risk of miscarriages
FSHR	136435	c.2039G>A (p.Ser680Asn)	Higher sensitivity to FSH (in women)
USP9Y AZFa, b, c	400005		Microdeletion AZF; Failure of spermatogenesis (in men)