Gene Variants Associated with Reproduction Disorders V4

- list of examined disORDERs

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| **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) |