

# UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS

## PANEL<sup>1</sup> DG-4.2.0 (29 GENES)

| <i>Gene</i> | <i>Twist X2 covered 10x</i> | <i>Twist X2 covered 20x</i> | <i>srWGS covered 10x</i> | <i>srWGS covered 15x</i> | <i>srWGS covered 20x</i> | <i>Associated Phenotype description and OMIM disease ID</i>                        |
|-------------|-----------------------------|-----------------------------|--------------------------|--------------------------|--------------------------|--|
| ASTL        | 100%                        | 100%                        | 100%                     | 100%                     | 99.5%                    | ?Oocyte/zygote/embryo maturation arrest 11, 619643                                 |
| BTG4        | 100%                        | 100%                        | 100%                     | 100%                     | 99.5%                    | Oocyte/zygote/embryo maturation arrest 8, 619009                                   |
| C11orf80    | 92.2%                       | 92.2%                       | 100%                     | 100%                     | 99.2%                    |  |
| CCNB3       | 100%                        | 100%                        | 99.2%                    | 91.2%                    | 71.5%                    |  |
| CDC20       | 100%                        | 100%                        | 100%                     | 100%                     | 98.8%                    | Oocyte/zygote/embryo maturation arrest 14, 620276                                  |
| FBXO43      | 100%                        | 100%                        | 100%                     | 100%                     | 99.6%                    | Spermatogenic failure 64, 619696;Oocyte/zygote/embryo maturation arrest 12, 619697 |
| KHDC3L      | 100%                        | 100%                        | 100%                     | 99.8%                    | 99.2%                    | Hydatidiform mole, recurrent, 2, 614293  |

|       |      |      |      |       |       |  |
|-------|------|------|------|-------|-------|--|
| KPNA7 | 100% | 100% | 100% | 100%  | 98.9% | Oocyte/zygote/embryo maturation arrest 17, 620319  |
| LHCGR | 100% | 100% | 100% | 99.9% | 99.1% | Leydig cell adenoma, somatic, with precocious puberty, 176410;Leydig cell hypoplasia with pseudohermaphroditism, 238320;Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320;Luteinizing hormone resistance, female, 238320;Precocious puberty, male, 176410 |
| MEI1  | 100% | 100% | 100% | 100%  | 99.3% | Hydatidiform mole, recurrent, 3, 618431  |
| MOS   | 100% | 100% | 100% | 99.4% | 97.1% | Oocyte/zygote/embryo maturation arrest 20, 620383  |
| NLRP2 | 100% | 100% | 100% | 100%  | 99.1% | Oocyte/zygote/embryo maturation arrest 18, 620332  |
| NLRP5 | 100% | 100% | 100% | 100%  | 99.1% | Oocyte/zygote/embryo maturation arrest 19, 620333  |
| NLRP7 | 100% | 100% | 100% | 99.9% | 99.2% | Hydatidiform mole, recurrent, 1, 231090  |
| OOEP  | 100% | 100% | 100% | 100%  | 99%   |  |

|         |      |       |       |       |       |  |
|---------|------|-------|-------|-------|-------|--|
| PABPC1L | 100% | 100%  | 100%  | 99.8% | 98.4% |  |
| PADI6   | 100% | 99.9% | 99.4% | 98.1% | 95.7% | Oocyte/zygote/embryo maturation arrest 16, 617234  |
| PANX1   | 100% | 100%  | 100%  | 100%  | 99.3% | Oocyte/zygote/embryo maturation arrest 7, 618550   |
| PATL2   | 100% | 100%  | 100%  | 99.9% | 99.3% | Oocyte/zygote/embryo maturation arrest 4, 617743   |
| REC114  | 100% | 100%  | 100%  | 99.9% | 99.1% | Oocyte/zygote/embryo maturation arrest 10, 619176  |
| TBPL2   | 100% | 100%  | 100%  | 100%  | 99.3% |  |
| TLE6    | 100% | 100%  | 100%  | 100%  | 98.3% | Oocyte/zygote/embryo maturation arrest 15, 616814  |
| TRIP13  | 100% | 100%  | 100%  | 99.9% | 99.1% | Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598 |
| TUBB8   | 100% | 100%  | 100%  | 100%  | 99.7% | Oocyte/zygote/embryo maturation arrest 2, 616780   |
| WEE2    | 100% | 100%  | 100%  | 100%  | 99.5% | Oocyte/zygote/embryo maturation arrest 5, 617996   |
| ZFP36L2 | 100% | 100%  | 100%  | 99.7% | 98.3% | Oocyte/zygote/embryo maturation arrest 13, 620154  |

|     |      |      |      |       |       |  |
|-----|------|------|------|-------|-------|--|
| ZP1 | 100% | 100% | 100% | 99.9% | 98.9% | Oocyte/zygote/embryo maturation arrest 1, 615774 |
| ZP2 | 100% | 100% | 100% | 100%  | 99.2% | Oocyte/zygote/embryo maturation arrest 6, 618353 |
| ZP3 | 100% | 100% | 100% | 100%  | 99.2% | Oocyte/zygote/embryo maturation arrest 3, 617712 |

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene’s coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

TWIST X2 covered 20x describes the percentage of a gene’s coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

srWGS covered 10x describes the percentage of a gene’s coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.

srWGS covered 15x describes the percentage of a gene’s coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.

srWGS covered 20x describes the percentage of a gene’s coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.

non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.

This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors