# PREMATURE OVARIAN INSUFFICIENCY PANEL DG-3.9.0 (46 GENES)

| *Gene* | *Twist X2 covered >10x* | *Twist X2 covered >20x* | *WGS covered >10x* | *WGS covered >20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|---|
| AARS2 | 100.0% | 100.0% | 100.0% | 99.4% | Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096 |
| BMP15 | 100.0% | 100.0% | 98.7% | 73.4% | Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510 |
| BNC1 | 100.0% | 99.9% | 100.0% | 98.4% | ?Premature ovarian failure 16, 618723 |
| C14orf39 | 100.0% | 100.0% | 100.0% | 96.6% | Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203 |
| CCDC155 | 100.0% | 100.0% | 100.0% | 98.8% | Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548 |
| CLPP | 100.0% | 100.0% | 100.0% | 96.3% | Perrault syndrome 3, 614129 |

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| CYP17A1 | 100.0% | 100.0% | 100.0% | 99.2% | 17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110 |
| CYP19A1 | 100.0% | 99.9% | 100.0% | 98.8% | Aromatase deficiency, 613546;Aromatase excess syndrome, 139300 |
| DCAF17 | 100.0% | 100.0% | 99.9% | 98.3% | Woodhouse-Sakati syndrome, 241080 |
| EIF2B5 | 100.0% | 100.0% | 100.0% | 98.9% | Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 |
| EIF4ENIF1 | 100.0% | 100.0% | 100.0% | 98.6% | |
| ERAL1 | 100.0% | 100.0% | 100.0% | 98.3% | Perrault syndrome 6, 617565 |
| ERCC6 | 100.0% | 100.0% | 100.0% | 98.8% | UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980 |
| ESR2 | 100.0% | 100.0% | 100.0% | 98.9% | ?Ovarian dysgenesis 8, 618187 |

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| FANCM | 100.0% | 100.0% | 100.0% | 97.3% | ?Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086 |
| FIGLA | 100.0% | 100.0% | 100.0% | 99.1% | Premature ovarian failure 6, 612310 |
| FIGNL1 | 100.0% | 100.0% | 100.0% | 99.2% | |
| FOXL2 | 100.0% | 100.0% | 99.8% | 88.9% | Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100;Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100;Premature ovarian failure 3, 608996 |
| FSHB | 98.7% | 98.0% | 100.0% | 99.8% | Hypogonadotropic hypogonadism 24 without anosmia, 229070 |
| FSHR | 100.0% | 99.9% | 100.0% | 99.3% | Ovarian response to FSH stimulation, 276400;Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300 |
| GALT | 100.0% | 100.0% | 100.0% | 99.2% | Galactosemia, 230400 |
| GDF9 | 100.0% | 100.0% | 100.0% | 98.9% | ?Premature ovarian failure 14, 618014 |
| GGPS1 | 100.0% | 100.0% | 100.0% | 98.7% | Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518 |
| HARS2 | 100.0% | 100.0% | 100.0% | 98.9% | Perrault syndrome 2, 614926 |

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| HFM1 | 100.0% | 100.0% | 100.0% | 96.4% | Premature ovarian failure 9, 615724 |
| HROB | 100.0% | 100.0% | 100.0% | 99.2% | |
| HSD17B4 | 96.6% | 96.6% | 100.0% | 98.2% | D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400 |
| HSF2BP | 100.0% | 100.0% | 100.0% | 98.4% | Premature ovarian failure 19, 619245 |
| LARS2 | 100.0% | 100.0% | 100.0% | 99.1% | Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021 |
| MCM8 | 94.4% | 94.4% | 100.0% | 98.8% | ?Premature ovarian failure 10, 612885 |
| MCM9 | 100.0% | 100.0% | 100.0% | 98.3% | Ovarian dysgenesis 4, 616185 |
| MSH4 | 100.0% | 100.0% | 100.0% | 98.3% | Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420 |
| NOBOX | 100.0% | 100.0% | 100.0% | 99.1% | Premature ovarian failure 5, 611548 |
| NR5A1 | 100.0% | 100.0% | 100.0% | 98.6% | 46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957 |

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| PMM2 | 100.0% | 100.0% | 100.0% | 98.1% | Congenital disorder of glycosylation, type Ia, 212065 |
| POLG | 100.0% | 100.0% | 100.0% | 99.4% | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450 |
| PSMC3IP | 100.0% | 100.0% | 100.0% | 99.0% | Ovarian dysgenesis 3, 614324 |
| SOHLH1 | 100.0% | 100.0% | 100.0% | 99.4% | Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115 |
| SOX11 | 100.0% | 100.0% | 100.0% | 90.9% | Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866 |
| SPATA22 | 100.0% | 100.0% | 100.0% | 98.0% | |
| SPIDR | 100.0% | 100.0% | 100.0% | 98.7% | Ovarian dysgenesis 9, 619665 |

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| STAG3 | 100.0% | 100.0% | 100.0% | 98.5% | Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723 |
| SYCE1 | 100.0% | 100.0% | 100.0% | 99.3% | ?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947 |
| TP63 | 100.0% | 99.9% | 100.0% | 99.3% | Premature ovarian failure 21, 620311;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292;Hay-Wells syndrome, 106260;Split-hand/foot malformation 4, 605289;Orofacial cleft 8, 618149;Rapp-Hodgkin syndrome, 129400;ADULT syndrome, 103285;Limb-mammary syndrome, 603543 |
| TWNK | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138 |
| ZNF541 | 100.0% | 100.0% | 100.0% | 98.9% | |

*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.*
*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.*

*srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.*
*This list is accurate for panel version DG 3.9.0*