PREMATURE OVARIAN INSUFFICIENCY PANEL DG-5.0.0 (55 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
AARS2	100%	100%	100%	99.9%	98.9%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ANKRD31	100%	100%	100%	99.9%	99.5%	
BMP15	100%	100%	99.2%	89.7%	69.7%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510
BNC1	100%	100%	100%	100%	99.7%	?Premature ovarian failure 16, 618723
C14orf39	100%	100%	100%	100%	99.6%	Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203
CLPP	100%	100%	100%	100%	99%	Perrault syndrome 3, 614129
CYP17A1	100%	100%	100%	100%	99.4%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	100%	100%	99.8%	Aromatase deficiency, 613546
DAP3	93.3%	91.2%	100%	100%	99.6%	Perrault syndrome 7, 621101
DCAF17	96.5%	96.5%	100%	100%	99.7%	Woodhouse-Sakati syndrome, 241080

EIF2B5	100%	100%	100%	100%	99.5%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF4ENIF1	100%	100%	100%	100%	99.5%	
ERAL1	100%	100%	100%	100%	99.9%	Perrault syndrome 6, 617565
ERCC6	100%	100%	100%	100%	99.5%	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
ESR1	100%	100%	100%	100%	99.4%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446
ESR2	100%	100%	100%	100%	99.5%	?Ovarian dysgenesis 8, 618187
FANCM	100%	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FIGLA	100%	100%	100%	100%	99.6%	Premature ovarian failure 6, 612310
FIGNL1	100%	100%	100%	100%	99.8%	

FOXL2	100%	100%	99.9%	99.5%	96.2%	Premature ovarian failure 3, 608996;Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2, 110100
FSHB	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	100%	100%	99.7%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
GALT	100%	100%	100%	99.9%	99.2%	Galactosemia, 230400
GDF9	100%	100%	100%	100%	99.9%	Premature ovarian failure 14, 618014
GGPS1	100%	100%	100%	100%	100%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
HARS2	100%	100%	100%	100%	99.6%	Perrault syndrome 2, 614926
HFM1	100%	100%	100%	100%	99.8%	Premature ovarian failure 9, 615724
HROB	95.2%	95.2%	100%	100%	99.6%	Ovarian dysgenesis 11, 620897
HSD17B4	100%	100%	100%	100%	99.8%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSF2BP	100%	100%	100%	100%	99.8%	Premature ovarian failure 19, 619245
KASH5	100%	100%	100%	100%	99.5%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548

LARS2	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHCGR	100%	100%	100%	100%	99.9%	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
MCM8	95.1%	94.4%	100%	100%	99.6%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	99.9%	99.5%	Ovarian dysgenesis 4, 616185
MEI4	100%	100%	100%	100%	99.8%	
MSH4	100%	100%	100%	100%	99.8%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
NOBOX	100%	100%	100%	100%	99.2%	Premature ovarian failure 5, 611548
NR5A1	99.1%	95.4%	100%	100%	99.2%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
PMM2	94.6%	94.6%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ia, 212065

POLG	100%	100%	100%	100%	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%	100%	100%	100%	99.9%	Ovarian dysgenesis 3, 614324
RNF111	97.5%	97.5%	100%	100%	99.6%	
SOHLH1	100%	100%	100%	100%	99.5%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX11	100%	100%	100%	99.5%	94.3%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SPATA22	100%	100%	100%	100%	99.9%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPIDR	100%	100%	100%	100%	99.4%	Ovarian dysgenesis 9, 619665
STAG3	100%	100%	100%	100%	99.2%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
SWSAP1	100%	100%	100%	100%	99.5%	

SYCE1	100%	100%	100%	100%	99.3%	?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947
SYCP2L	100%	100%	100%	100%	99.7%	Premature ovarian failure 24, 620840
TP63	100%	100%	100%	100%	99.7%	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%	100%	100%	100%	99.7%	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%	100%	100%	100%	99.5%	
ZSWIM7	94.6%	88.6%	100%	99.9%	99.4%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834

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 4.4.0

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