PREMATURE OVARIAN INSUFFICIENCY PANEL DG-4.1.0 (48 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%	100%	100%	99.3%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
BMP15	100%	99.7%	98.8%	72.8%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510
BNC1	100%	99.7%	100%	99.6%	?Premature ovarian failure 16, 618723
C14orf39	100%	100%	100%	99.8%	Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203
CLPP	100%	100%	100%	96%	Perrault syndrome 3, 614129
CYP17A1	100%	100%	100%	98.7%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110

CYP19A1	100%	100%	100%	99.8%	Aromatase deficiency, 613546;Aromatase excess syndrome, 139300
DCAF17	100%	100%	100%	99.4%	Woodhouse-Sakati syndrome, 241080
EIF2B5	100%	100%	100%	99.4%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF4ENIF1	100%	100%	100%	99.4%	
ERAL1	100%	99.9%	100%	99.6%	Perrault syndrome 6, 617565
ERCC6	100%	100%	100%	99.5%	UV-sensitive syndrome 1, 600630;Cerebrooculofacios keletal 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%	100%	100%	99.5%	?Ovarian dysgenesis 8, 618187
FANCM	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086

FIGLA	100%	100%	100%	99.6%	Premature ovarian failure 6, 612310
FIGNL1	100%	100%	100%	99.8%	
FOXL2	100%	100%	99.9%	91.8%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100;Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100;Premature ovarian failure 3, 608996
FSHB	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	100%	99.7%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
GALT	100%	100%	100%	99%	Galactosemia, 230400
GDF9	100%	100%	100%	99.6%	Premature ovarian failure 14, 618014
GGPS1	100%	100%	100%	99.7%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
HARS2	100%	100%	100%	99.2%	Perrault syndrome 2, 614926
HFM1	100%	100%	100%	99.7%	Premature ovarian failure 9, 615724
HROB	100%	100%	100%	99%	Ovarian dysgenesis 11, 620897

HSD17B4	100%	100%	100%	99.6%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSF2BP	100%	100%	100%	99.5%	Premature ovarian failure 19, 619245
KASH5	100%	100%	100%	98.8%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
LARS2	100%	100%	100%	99.6%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
MCM8	94.4%	94.4%	100%	99.8%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	99.5%	Ovarian dysgenesis 4, 616185
MSH4	100%	100%	100%	99.7%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
NOBOX	100%	100%	100%	98.6%	Premature ovarian failure 5, 611548
NR5A1	100%	99.9%	100%	98.9%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957

PMM2	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
POLG	100%	100%	100%	99.5%	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%	99.8%	Ovarian dysgenesis 3, 614324
RNF111	100%	100%	100%	99.3%	
SOHLH1	100%	100%	100%	98.6%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX11	100%	100%	100%	94.3%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866

SPATA22	100%	100%	100%	99.9%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPIDR	100%	100%	100%	99.4%	Ovarian dysgenesis 9, 619665
STAG3	100%	100%	100%	99.1%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
SYCE1	100%	100%	100%	99.1%	?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947
SYCP2L	100%	100%	100%	99.6%	Premature ovarian failure 24, 620840
TP63	100%	100%	100%	99.6%	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%	99.5%	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%	99.2%	

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 4.0.0

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors