PREMATURE OVARIAN INSUFFICIENCY PANEL DG-5.2.0 (55 GENES)

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
AARS2	100% | 100%	97% | 97%	99% | 99%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ANKRD31	100% | 100%	98% | 98%	99% | 99%	
BMP15	99% | 99%	99% | 99%	100% | 100%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510
BNC1	100% | 100%	97% | 97%	99% | 99%	?Premature ovarian failure 16, 618723
C14orf39	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 52, 619202;Premature ovarian failure 18, 619203
CLPP	100% | 100%	96% | 96%	98% | 98%	Perrault syndrome 3, 614129
CYP17A1	100% | 100%	96% | 96%	99% | 99%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100% | 100%	98% | 98%	100% | 100%	Aromatase deficiency, 613546
DAP3	91% | 91%	97% | 97%	99% | 99%	Perrault syndrome 7, 621101
DCAF17	97% | 97%	97% | 97%	99% | 99%	Woodhouse-Sakati syndrome, 241080
EIF2B5	100% | 100%	97% | 97%	99% | 99%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF4ENIF1	100% | 100%	96% | 96%	99% | 99%	
ERAL1	99% | 99%	97% | 97%	99% | 99%	Perrault syndrome 6, 617565
ERCC6	100% | 100%	98% | 98%	99% | 99%	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%	96% | 96%	99% | 99%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446

ESR2	100% | 100%	97% | 97%	99% | 99%	?Ovarian dysgenesis 8, 618187
FANCM	100% | 100%	98% | 98%	100% | 100%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FIGLA	99% | 99%	96% | 96%	99% | 99%	Premature ovarian failure 6, 612310
FIGNL1	100% | 100%	98% | 98%	99% | 99%	
FOXL2	100% | 100%	91% | 91%	99% | 99%	Premature ovarian failure 3, 608996;Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2, 110100
FSHB	100% | 100%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100% | 100%	97% | 97%	99% | 99%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
GALT	100% | 100%	97% | 97%	99% | 99%	Galactosemia, 230400
GDF9	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 14, 618014
GGPS1	100% | 100%	98% | 98%	99% | 99%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
HARS2	100% | 100%	97% | 97%	99% | 99%	Perrault syndrome 2, 614926
HFM1	100% | 100%	98% | 98%	99% | 99%	?Hydatidiform mole, recurrent, 5, 621720;Premature ovarian failure 9, 615724
HROB	96% | 96%	97% | 97%	99% | 99%	Ovarian dysgenesis 11, 620897
HSD17B4	100% | 100%	96% | 96%	100% | 100%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSF2BP	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 19, 619245
KASH5	100% | 100%	97% | 96%	98% | 98%	Hydatidiform mole, recurrent, 6, 621721;Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
LARS2	96% | 96%	97% | 97%	99% | 99%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LHCGR	100% | 100%	98% | 98%	99% | 99%	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	94% | 94%	98% | 98%	100% | 100%	?Premature ovarian failure 10, 612885
MCM9	100% | 100%	97% | 97%	99% | 99%	Ovarian dysgenesis 4, 616185
MEI4	100% | 100%	98% | 98%	100% | 100%	
MSH4	100% | 100%	98% | 98%	99% | 99%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
NOBOX	100% | 100%	96% | 96%	97% | 97%	Premature ovarian failure 5, 611548
NR5A1	94% | 93%	96% | 96%	98% | 98%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957

PMM2	95% | 95%	97% | 97%	99% | 99%	Congenital disorder of glycosylation, type Ia, 212065
POLG	100% | 100%	97% | 97%	99% | 99%	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%	97% | 97%	99% | 99%	Ovarian dysgenesis 3, 614324
RNF111	98% | 98%	97% | 97%	99% | 99%	
SOHLH1	99% | 99%	95% | 95%	97% | 97%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX11	100% | 100%	93% | 93%	99% | 99%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SPATA22	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPIDR	100% | 100%	97% | 97%	99% | 99%	Ovarian dysgenesis 9, 619665
STAG3	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
SWSAP1	100% | 100%	97% | 97%	98% | 98%	
SYCE1	99% | 99%	96% | 96%	99% | 99%	Premature ovarian failure 12, 616947;Spermatogenic failure 15, 616950
SYCP2L	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 24, 620840
TP63	100% | 100%	98% | 98%	100% | 100%	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%	97% | 97%	99% | 99%	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%	97% | 97%	98% | 98%	
ZSWIM7	85% | 83%	97% | 97%	99% | 99%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834