PRIMARY OVARIAN INSUFFICIENCY GENE PANEL DG 3.3.0 (29 genes)

Releasedate: 13-01-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
AARS2	100%	100%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
BMP15	100%	100%	Premature ovarian failure 4, 300510 Ovarian dysgenesis 2, 300510
CLPP	100%	100%	Perrault syndrome 3, 614129
CYP17A1	100%	100%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
EIF2B5	100%	100%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
ERAL1	100%	100%	Perrault syndrome 6, 617565
ESR2	100%	100%	?Ovarian dysgenesis 8, 618187
FANCM	100%	100%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FOXL2	100%	100%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Premature ovarian failure 3, 608996
FSHB	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300
GALT	100%	100%	Galactosemia, 230400
GDF9	100%	100%	?Premature ovarian failure 14, 618014
HARS2	100%	100%	Perrault syndrome 2, 614926
HFM1	100%	100%	Premature ovarian failure 9, 615724
HSD17B4	96%	96%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400

LARS2	100%	100%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
MCM8	94%	94%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	Ovarian dysgenesis 4, 616185
MSH4	100%	100%	No OMIM disease ID
NOBOX	100%	100%	Premature ovarian failure 5, 611548
NR5A1	100%	100%	46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Spermatogenic failure 8, 613957
PMM2	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
POLG	100%	100%	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%	Ovarian dysgenesis 3, 614324
SOHLH1	100%	100%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
STAG3	100%	100%	Premature ovarian failure 8, 615723
TWNK	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138

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.
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TWIST is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 : January 13th , 2022.
This list is accurate for panel version DG 3.3.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors