

Gene panel information

Gene panel	OZEMA-TFF
Version	4
Total genes	72
Activation date	Thursday 01 october 2026
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ACR	96.52 %	102480	?Spermatogenic failure 87, 620500 (3), Autosomal recessive
ACTL7A	99.99 %	604303	Spermatogenic failure 86, 620499 (3), Autosomal recessive
ACTL9	99.78 %	619251	Spermatogenic failure 53, 619258 (3), Autosomal recessive
ANAPC13	99.79 %	614484	No OMIM phenotypes
ASTL	99.08 %	608860	?Oocyte/zygote/embryo maturation arrest 11, 619643 (3), Autosomal recessive
BTG4	99.92 %	605673	Oocyte/zygote/embryo maturation arrest 8, 619009 (3), Autosomal recessive
BUB1B	99.99 %	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
C11orf80	99.41 %	616109	Hydatidiform mole, recurrent, 4, 618432 (3), Autosomal recessive
C2CD6	99.6 %	619776	?Spermatogenic failure 68, 619805 (3), Autosomal recessive
CATSPER3	99.92 %	609120	No OMIM phenotypes
CCDC174	99.98 %	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
CCDC62	99.93 %	613481	?Spermatogenic failure 67, 619803 (3), Autosomal recessive
CCIN	99.99 %	603960	Spermatogenic failure 91, 620838 (3), Autosomal recessive
CCNB3	99.91 %	300456	No OMIM phenotypes
CDC20	99.44 %	603618	Oocyte/zygote/embryo maturation arrest 14, 620276 (3), Autosomal recessive
CDC23	99.76 %	603462	No OMIM phenotypes
CDC26	93.97 %	614533	No OMIM phenotypes
CENPH	99.83 %	605607	No OMIM phenotypes
CHEK1	99.67 %	603078	Oocyte/zygote/embryo maturation arrest 21, 620610 (3), Autosomal dominant
CIP2A	99.48 %	610643	No OMIM phenotypes
COX15	99.92 %	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
DLGAP5	99.87 %	617859	No OMIM phenotypes
DNAH17	99.85 %	610063	Spermatogenic failure 39, 618643 (3), Autosomal recessive
DPY19L2	91.21 %	613893	Spermatogenic failure 9, 613958 (3), Autosomal recessive
ELL3	99.75 %	609885	No OMIM phenotypes
FBXO43	99.92 %	609110	Spermatogenic failure 64, 619696 (3), Autosomal recessive; Oocyte/zygote/embryo maturation arrest 12, 619697 (3), Autosomal recessive
GGN	99.62 %	609966	Spermatogenic failure 69, 619826 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GMNN	99.29 %	602842	Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant
HAUS6	99.38 %	613433	No OMIM phenotypes
HFM1	95.66 %	615684	Premature ovarian failure 9, 615724 (3), Autosomal recessive
IQCN	99.98 %	620160	Spermatogenic failure 78, 620170 (3), Autosomal recessive
JARID2	99.88 %	601594	Developmental delay with variable intellectual disability and dysmorphic facies, 620098 (3), Autosomal dominant
KCNU1	99.65 %	615215	Spermatogenic failure 79, 620196 (3), Autosomal recessive
KHDC3L	99.99 %	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
KIF11	99.79 %	148760	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950 (3), Autosomal dominant
KIF18A	99.87 %	611271	No OMIM phenotypes
KPNA7	99.13 %	614107	Oocyte/zygote/embryo maturation arrest 17, 620319 (3), Autosomal recessive
MAD2L1BP	96 %	618136	No OMIM phenotypes
MEI1	99.99 %	608797	Hydatidiform mole, recurrent, 3, 618431 (3), Autosomal recessive
MLH3	99.98 %	604395	{Endometrial cancer, susceptibility to}, 608089 (3), Somatic mutation, Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Colorectal cancer, hereditary nonpolyposis, type 7, 614385 (3)
MOS	99.91 %	190060	Oocyte/zygote/embryo maturation arrest 20, 620383 (3), Autosomal recessive
NLRP14	99.94 %	609665	No OMIM phenotypes
NLRP2	99.99 %	609364	Oocyte/zygote/embryo maturation arrest 18, 620332 (3), Autosomal recessive
NLRP5	99.92 %	609658	Oocyte/zygote/embryo maturation arrest 19, 620333 (3), Autosomal recessive
NLRP7	99.9 %	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
OOEP	99.97 %	611689	No OMIM phenotypes
PABPC1L	99.47 %	621055	Oocyte/zygote/embryo maturation arrest 22, 621093 (3), Autosomal recessive
PADI6	98.74 %	610363	Oocyte/zygote/embryo maturation arrest 16, 617234 (3), Autosomal recessive
PANX1	99.96 %	608420	Oocyte/zygote/embryo maturation arrest 7, 618550 (3), Autosomal dominant
PATL2	99.92 %	614661	Oocyte/zygote/embryo maturation arrest 4, 617743 (3), Autosomal recessive
PICK1	99.97 %	605926	No OMIM phenotypes
PLCZ1	99.83 %	608075	Spermatogenic failure 17, 617214 (3), Autosomal recessive
REC114	99.6 %	618421	Oocyte/zygote/embryo maturation arrest 10, 619176 (3), Autosomal recessive
SEPTIN12	99.6 %	611562	Spermatogenic failure 10, 614822 (3), Autosomal dominant
SPATA16	99.9 %	609856	?Spermatogenic failure 6, 102530 (3), Autosomal recessive
SPINK2	99.93 %	605753	?Spermatogenic failure 29, 618091 (3), Autosomal recessive
TACC3	99.97 %	605303	No OMIM phenotypes
TBPL2	99.96 %	608964	No OMIM phenotypes
TLE6	99.93 %	612399	Oocyte/zygote/embryo maturation arrest 15, 616814 (3), Autosomal recessive
TRIP13	99.98 %	604507	Oocyte/zygote/embryo maturation arrest 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TUBA1C	99.73 %	621161	Oocyte/zygote/embryo maturation arrest 24, 621232 (3), Autosomal recessive
TUBA4A	100 %	191110	Oocyte/zygote/embryo maturation arrest 23, 621231 (3), Autosomal recessive, Autosomal dominant; Spastic ataxia 11, autosomal dominant, 621226 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 9, 616208 (3), Autosomal dominant; Congenital myopathy 26, 621225 (3), Autosomal dominant
TUBB8	99.19 %	616768	Oocyte/zygote/embryo maturation arrest 2, 616780 (3), Autosomal recessive, Autosomal dominant
WEE2	99.94 %	614084	Oocyte/zygote/embryo maturation arrest 5, 617996 (3), Autosomal recessive
YTHDC2	99.58 %	616530	<i>No OMIM phenotypes</i>
ZAR1	99.53 %	607520	<i>No OMIM phenotypes</i>
ZFP36L2	99.88 %	612053	Oocyte/zygote/embryo maturation arrest 13, 620154 (3), Autosomal recessive
ZP1	99.57 %	195000	Oocyte/zygote/embryo maturation arrest 1, 615774 (3), Autosomal recessive
ZP2	99.53 %	182888	Oocyte/zygote/embryo maturation arrest 6, 618353 (3), Autosomal recessive
ZP3	90.56 %	182889	Oocyte/zygote/embryo maturation arrest 3, 617712 (3), Autosomal dominant
ZP4	99.92 %	613514	<i>No OMIM phenotypes</i>
ZPBP	98.39 %	608498	?Spermatogenic failure 66, 619799 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

- (1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
- (2) the disorder has been placed on the map by linkage; no mutation has been found
- (3) the molecular basis for the disorder is known; a mutation has been found in the gene
- (4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.