Fertilisation failure-oocyte maturation arrest-embryonic arrest

Gene panel

Gene panel information

Gene panel	**Fertilisation failure-oocyte maturation arrest-embryonic arrest**
Version	3
Total genes	51
Activation date	Wednesday 13 march 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
ACR	97.24 %	102480	?Male infertility due to acrosin deficiency, 102480 (2)
ACTL7A	100 %	604303	*No OMIM phenotypes*
ACTL9	100 %	619251	Spermatogenic failure 53, 619258 (3), Autosomal recessive
ASTL	99.39 %	608860	?Oocyte/zygote/embryo maturation arrest 11, 619643 (3), Autosomal recessive
BTG4	99.98 %	605673	Oocyte/zygote/embryo maturation arrest 8, 619009 (3), Autosomal recessive
BUB1B	100 %	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
C11orf80	99.89 %	616109	Hydatidiform mole, recurrent, 4, 618432 (3), Autosomal recessive
C2CD6	99.29 %	619776	?Spermatogenic failure 68, 619805 (3), Autosomal recessive
CATSPER3	99.98 %	609120	*No OMIM phenotypes*
CCDC62	99.98 %	613481	?Spermatogenic failure 67, 619803 (3), Autosomal recessive
CCIN	99.99 %	603960	*No OMIM phenotypes*
CCNB3	99.96 %	300456	*No OMIM phenotypes*
CDC20	99.89 %	603618	Oocyte/zygote/embryo maturation arrest 14, 620276 (3), Autosomal recessive
CHEK1	99.89 %	603078	*No OMIM phenotypes*
DNAH17	99.98 %	610063	Spermatogenic failure 39, 618643 (3), Autosomal recessive
DPY19L2	92.02 %	613893	Spermatogenic failure 9, 613958 (3), Autosomal recessive
FBXO43	99.96 %	609110	Spermatogenic failure 64, 619696 (3), Autosomal recessive; Oocyte/zygote/embryo maturation arrest 12, 619697 (3), Autosomal recessive
GGN	99.99 %	609966	Spermatogenic failure 69, 619826 (3), Autosomal recessive
IQCN	100 %	620160	Spermatogenic failure 78, 620170 (3), Autosomal recessive
KCNU1	99.93 %	615215	Spermatogenic failure 79, 620196 (3), Autosomal recessive
KHDC3L	100 %	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
KPNA7	99.51 %	614107	Oocyte/zygote/embryo maturation arrest 17, 620319 (3), Autosomal recessive
MEI1	99.99 %	608797	Hydatidiform mole, recurrent, 3, 618431 (3), Autosomal recessive
MOS	100 %	190060	Oocyte/zygote/embryo maturation arrest 20, 620383 (3), Autosomal recessive
NLRP2	99.99 %	609364	Oocyte/zygote/embryo maturation arrest 18, 620332 (3), Autosomal recessive
NLRP5	100 %	609658	Oocyte/zygote/embryo maturation arrest 19, 620333 (3), Autosomal recessive
NLRP7	99.99 %	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
OOEP	99.97 %	611689	*No OMIM phenotypes*
PABPC1L	99.96 %		*No OMIM phenotypes*

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PADI6	99.47 %	610363	Oocyte/zygote/embryo maturation arrest 16, 617234 (3), Autosomal recessive
PANX1	100 %	608420	Oocyte/zygote/embryo maturation arrest 7, 618550 (3), Autosomal dominant
PATL2	99.99 %	614661	Oocyte/zygote/embryo maturation arrest 4, 617743 (3), Autosomal recessive
PICK1	100 %	605926	*No OMIM phenotypes*
PLCZ1	99.83 %	608075	Spermatogenic failure 17, 617214 (3), Autosomal recessive
REC114	99.89 %	618421	Oocyte/zygote/embryo maturation arrest 10, 619176 (3), Autosomal recessive
RGS12	99.99 %	602512	*No OMIM phenotypes*
SEPTIN12	99.94 %	611562	Spermatogenic failure 10, 614822 (3), Autosomal dominant
SPATA16	99.96 %	609856	?Spermatogenic failure 6, 102530 (3), Autosomal recessive
SPINK2	100 %	605753	?Spermatogenic failure 29, 618091 (3), Autosomal recessive
TBPL2	99.96 %	608964	*No OMIM phenotypes*
TLE6	100 %	612399	Oocyte/zygote/embryo maturation arrest 15, 616814 (3), Autosomal recessive
TRIP13	100 %	604507	Oocyte/zygote/embryo maturation arrest 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive
TUBA4A	100 %	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant
TUBB8	99.83 %	616768	Oocyte/zygote/embryo maturation arrest 2, 616780 (3), Autosomal recessive, Autosomal dominant
WEE2	99.98 %	614084	Oocyte/zygote/embryo maturation arrest 5, 617996 (3), Autosomal recessive
ZAR1	99.99 %	607520	*No OMIM phenotypes*
ZFP36L2	100 %	612053	Oocyte/zygote/embryo maturation arrest 13, 620154 (3), Autosomal recessive
ZP1	99.94 %	195000	Oocyte/zygote/embryo maturation arrest 1, 615774 (3), Autosomal recessive
ZP2	99.79 %	182888	Oocyte/zygote/embryo maturation arrest 6, 618353 (3), Autosomal recessive
ZP3	93.11 %	182889	Oocyte/zygote/embryo maturation arrest 3, 617712 (3), Autosomal dominant
ZPBP	99.08 %	608498	?Spermatogenic failure 66, 619799 (3), Autosomal recessive

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Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

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.

* Exome panels: >=20x, HyperCap panels: >=30x