Subfertility-infertility-gamete malfunction panel

versie	v1 (34 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ACTL7A	604303	No OMIM phenotype
ACTL9	619251	Spermatogenic failure 53, 619258 (3), Autosomal recessive
BTG4	605673	Oocyte maturation defect 8, 619009 (3), Autosomal recessive
BUB1B	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
C11orf80	616109	Hydatidiform mole, recurrent, 4, 618432 (3), Autosomal recessive
CDC20	603618	No OMIM phenotype
CHEK1	603078	No OMIM phenotype
FBXO43	609110	Oocyte maturation defect 12, 619697 (3), Autosomal recessive; Spermatogenic failure 64, 619696 (3), Autosomal recessive
IQCN	No OMIM gene	No OMIM phenotype
KCNU1	615215	No OMIM phenotype
KHDC3L	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
MEI1	608797	Hydatidiform mole, recurrent, 3, 618431 (3), Autosomal recessive
MOS	190060	No OMIM phenotype
NLRP2	609364	No OMIM phenotype
NLRP5	609658	No OMIM phenotype
NLRP7	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
OOEP	611689	No OMIM phenotype
PADI6	610363	Preimplantation embryonic lethality 2, 617234 (3), Autosomal recessive
PANX1	608420	Oocyte maturation defect 7, 618550 (3), Autosomal dominant
PATL2	614661	Oocyte maturation defect 4, 617743 (3), Autosomal recessive
PLCZ1	608075	Spermatogenic failure 17, 617214 (3), Autosomal recessive
REC114	618421	Oocyte maturation defect 10, 619176 (3), Autosomal recessive
RGS12	602512	No OMIM phenotype
SEPTIN12	611562	Spermatogenic failure 10, 614822 (3), Autosomal dominant
TBPL2	608964	No OMIM phenotype
TLE6	612399	Preimplantation embryonic lethality, 616814 (3), Autosomal recessive
TRIP13	604507	Oocyte maturation defect 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive
TUBB8	616768	Oocyte maturation defect 2, 616780 (3), Autosomal dominant, Autosomal recessive
WEE2	614084	Oocyte maturation defect 5, 617996 (3), Autosomal recessive
ZAR1	607520	No OMIM phenotype
ZFP36L2	612053	No OMIM phenotype
ZP1	195000	Oocyte maturation defect 1, 615774 (3), Autosomal recessive

ZP2	182888	Oocyte maturation defect 6, 618353 (3), Autosomal recessive
ZP3	182889	Oocyte maturation defect 3, 617712 (3), Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: August 24, 2022

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.