MALE INFERTILITY GENE PANEL DG 3.5.0 (130 genes)

Releasedate: 05-12-2022

Gene	TWIST X2 covered >10x	TWIST X2 covered >20x	Associated Phenotype description and OMIM disease ID
ACTL9	100%	100%	Spermatogenic failure 53, 619258
ADAD2	100%	100%	No OMIM disease ID
ADCY10	100%	100%	No OMIM disease ID
ADGRG2	100%	100%	Congenital bilateral absence of vas deferens, X-linked, 300985
AR	100%	99%	Androgen insensitivity, partial, with or without breast cancer, 312300 Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Hypospadias 1, X-linked, 300633
ARMC2	100%	100%	Spermatogenic failure 38, 618433
AURKC	100%	100%	Spermatogenic failure 5, 243060
BPY2	50%	49%	No OMIM disease ID
BPY2B	50%	49%	No OMIM disease ID
BPY2C	50%	50%	No OMIM disease ID
C14orf39	100%	100%	Spermatogenic failure 52, 619202 ?Premature ovarian failure 18, 619203
CATIP	100%	100%	?Spermatogenic failure 54, 619379
CATSPER1	100%	100%	Spermatogenic failure 7, 612997
CATSPER2	100%	100%	No OMIM disease ID
CCDC155	100%	100%	No OMIM disease ID
CCDC39	100%	100%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	Ciliary dyskinesia, primary, 15, 613808
CDC14A	100%	100%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDY1	50%	50%	No OMIM disease ID
CDY1B	50%	50%	No OMIM disease ID

CDY2A	50%	50%	No OMIM disease ID
CDY2B	50%	50%	No OMIM disease ID
			Hydrocephalus, normal pressure, 1, 236690
CFAP43	100%	100%	Spermatogenic failure 19, 617592
CFAP44	100%	100%	Spermatogenic failure 20, 617593
CFAP58	100%	100%	Spermatogenic failure 49, 619144
CFAP65	100%	100%	Spermatogenic failure 40, 618664
CFAP69	100%	100%	Spermatogenic failure 24, 617959
			Cystic fibrosis, 219700
			Congenital bilateral absence of vas deferens, 277180
CFTR	100%	100%	Sweat chloride elevation without CF,
DAZ1	50%	50%	No OMIM disease ID
DAZ2	50%	50%	No OMIM disease ID
DAZ3	50%	49%	No OMIM disease ID
DAZ4	50%	49%	No OMIM disease ID
DCAF12L1	100%	100%	No OMIM disease ID
DDX3Y	50%	50%	No OMIM disease ID
DMC1	100%	100%	No OMIM disease ID
DMRT1	100%	100%	No OMIM disease ID
DNAAF2	100%	100%	Ciliary dyskinesia, primary, 10, 612518
DNAAF4	100%	100%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	100%	Ciliary dyskinesia, primary, 18, 614874
			Spermatogenic failure 18, 617576
DNAH1	100%	100%	?Ciliary dyskinesia, primary, 37, 617577
DNAH17	100%	100%	Spermatogenic failure 39, 618643
DNAH7	100%	100%	No OMIM disease ID
DNAH8	100%	100%	Spermatogenic failure 46, 619095
DNAI1	100%	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100%	100%	Ciliary dyskinesia, primary, 34, 617091
DNHD1	100%	100%	Spermatogenic failure 65, 619712
DPY19L2	100%	100%	Spermatogenic failure 9, 613958
E2F1	100%	100%	No OMIM disease ID

EIF1AY	50%	50%	No OMIM disease ID
FANCA	100%	100%	Fanconi anemia, complementation group A, 227650
FANCM	100%	100%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FKBP6	100%	100%	Spermatogenic failure 77, 620103
FSIP2	100%	100%	Spermatogenic failure 34, 618153
GALNTL5	100%	100%	No OMIM disease ID
GAS8	100%	100%	Ciliary dyskinesia, primary, 33, 616726
GCNA	100%	100%	Spermatogenic failure, X-linked, 4, 301077
HSF2	100%	100%	No OMIM disease ID
HSFY1	50%	50%	No OMIM disease ID
HSFY2	50%	49%	No OMIM disease ID
KDM5D	49%	49%	No OMIM disease ID
KLHL10	100%	100%	Spermatogenic failure 11, 615081
LRRC6	100%	100%	Ciliary dyskinesia, primary, 19, 614935
M1AP	100%	100%	Spermatogenic failure 48, 619108
MAATS1	100%	100%	Spermatogenic failure 51, 619177
MCM9	100%	100%	Ovarian dysgenesis 4, 616185
MEI1	100%	100%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100%	100%	?Spermatogenic failure 22, 617706
MLH3	100%	100%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MNS1	100%	100%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MSH4	100%	100%	Premature ovarian failure 20, 619938 Spermatogenic failure 2, 108420
MSH5	100%	100%	?Premature ovarian failure 13, 617442 Spermatogenic failure 74, 619937
NR0B1	100%	100%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018

			46, XX sex reversal 4, 617480
			Premature ovarian failure 7, 612964
			46XY sex reversal 3, 612965
			Adrenocortical insufficiency, 612964
NR5A1	100%	100%	Spermatogenic failure 8, 613957
PDHA2	100%	100%	Spermatogenic failure 70, 619828
PIH1D3	100%	100%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIWIL2	100%	100%	No OMIM disease ID
PKD1	100%	100%	Polycystic kidney disease 1, 173900
PLCZ1	100%	100%	Spermatogenic failure 17, 617214
PLK4	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PMFBP1	100%	100%	Spermatogenic failure 31, 618112
PNLDC1	100%	100%	Spermatogenic failure 57, 619528
PRY	50%	50%	No OMIM disease ID
PRY2	50%	50%	No OMIM disease ID
QRICH2	100%	100%	Spermatogenic failure 35, 618341
RAD21L1	100%	100%	No OMIM disease ID
RBBP7	100%	100%	No OMIM disease ID
RBMY1A1	50%	50%	No OMIM disease ID
RBMY1B	50%	50%	No OMIM disease ID
RBMY1D	50%	49%	No OMIM disease ID
RBMY1E	50%	50%	No OMIM disease ID
RBMY1F	49%	49%	No OMIM disease ID
RBMY1J	50%	50%	No OMIM disease ID
			?Spermatogenic failure 62, 619673
RNF212	100%	100%	Recombination rate QTL 1, 612042
RPS4Y2	50%	50%	No OMIM disease ID
RSPH3	100%	100%	Ciliary dyskinesia, primary, 32, 616481
RSPH9	100%	100%	Ciliary dyskinesia, primary, 12, 612650
SEPTIN12	100%	100%	Spermatogenic failure 10, 614822
SHOC1	100%	100%	Spermatogenic failure 75, 619949
SPAG6	100%	100%	No OMIM disease ID
SPATA16	100%	100%	?Spermatogenic failure 6, 102530

SPATA22		100%	100%	No OMIM disease ID
SPEF2		100%	100%	Spermatogenic failure 43, 618751
SPINK2		97%	97%	?Spermatogenic failure 29, 618091
SPO11		100%	100%	No OMIM disease ID
SRY		50%	50%	46XY sex reversal 1, 400044
STAG3		100%	100%	Spermatogenic failure 61, 619672 Premature ovarian failure 8, 615723
SUN5		100%	100%	Spermatogenic failure 16, 617187
SYCE1		100%	100%	?Spermatogenic failure 15, 616950 ?Premature ovarian failure 12, 616947
SYCP2		100%	100%	Spermatogenic failure 1, 258150
SYCP3		100%	100%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
TAF4B		100%	100%	?Spermatogenic failure 13, 615841
TDRD9		100%	100%	?Spermatogenic failure 30, 618110
TERB1		100%	100%	Spermatogenic failure 60, 619646
TERB2		100%	100%	?Spermatogenic failure 59, 619645
TEX11		97%	97%	Spermatogenic failure, X-linked 2, 309120
TEX14		100%	100%	Spermatogenic failure 23, 617707
TEX15		100%	100%	Spermatogenic failure 25, 617960
TRIM71		100%	100%	Hydrocephalus, congenital communicating, 1, 618667
TSGA10		100%	100%	?Spermatogenic failure 26, 617961
TTC29		100%	99%	Spermatogenic failure 42, 618745
UBR2		100%	100%	No OMIM disease ID
USP26		100%	100%	No OMIM disease ID
USP9Y		50%	50%	Spermatogenic failure, Y-linked, 2, 415000
WDR66		100%	100%	Spermatogenic failure 33, 618152
XKRY	NC		NC	No OMIM disease ID
XKRY2	NC		NC	No OMIM disease ID
XRCC2		100%	100%	Spermatogenic failure 50, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
ZFX		100%	100%	No OMIM disease ID

ZMYND15	100%	100%	?Spermatogenic failure 14, 615842

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 X2 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 : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors