MALE INFERTILITY PANEL DG-4.1.0 (187 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACTL7A	100%	100%	100%	99.1%	Spermatogenic failure 86, 620499
ACTL9	100%	100%	100%	99.4%	Spermatogenic failure 53, 619258
ACTRT1	100%	100%	98.6%	71.3%	
ADAD2	100%	100%	100%	99.1%	
ADCY10	100%	100%	100%	99.6%	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADGB	100%	100%	100%	99.3%	
ADGRG2	100%	99.7%	99%	74.2%	Congenital bilateral absence of vas deferens, X-linked, 300985
AK7	100%	100%	100%	99.6%	?Spermatogenic failure 27, 617965
AK9	100%	100%	100%	99.8%	Spermatogenic failure 89, 620705
AKAP3	100%	100%	100%	99.6%	Spermatogenic failure 82, 620353

AR	100%	99.6%	97.5%	67.6%	Androgen insensitivity, partial, with or without breast cancer, 312300;Spinal and bulbar muscular atrophy, X-linked 1, 313200;{Prostate cancer, susceptibility to}, 301120;Androgen insensitivity, 300068;Hypospadias 1, X-linked, 300633
ARMC2	100%	100%	100%	99.8%	Spermatogenic failure 38, 618433
AURKC	100%	100%	100%	98.8%	Spermatogenic failure 5, 243060
BCORL1	100%	99%	97.4%	65.1%	Shukla-Vernon syndrome, 301029
BNC1	100%	99.7%	100%	99.6%	?Premature ovarian failure 16, 618723
BPY2	50%	50%	49.2%	24.8%	
BPY2B	50%	50%	48.1%	27.8%	
BPY2C	50%	50%	48%	26.1%	
C14orf39	100%	100%	100%	99.8%	Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203
CALR3	100%	100%	100%	99.3%	
CATIP	100%	100%	100%	99.4%	?Spermatogenic failure 54, 619379
CATSPER1	100%	100%	100%	98.4%	Spermatogenic failure 7, 612997

CATSPER2	100%	100%	99.9%	96.6%	
CCDC146	100%	100%	100%	99.6%	Spermatogenic failure 94, 620850
CCDC34	100%	100%	100%	99.7%	Spermatogenic failure 76, 620084
CCDC39	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	100%	97.8%	Ciliary dyskinesia, primary, 15, 613808
CDC14A	100%	100%	100%	99.7%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDY1	50%	50%	47.1%	29.3%	
CDY1B	50%	50%	47.1%	27.1%	
CDY2A	50%	50%	49.6%	27%	
CDY2B	50%	50%	49.1%	25.7%	
CFAP251	100%	100%	100%	99.3%	Spermatogenic failure 33, 618152
CFAP43	100%	100%	100%	99.8%	Hydrocephalus, normal pressure, 1, 236690;Spermatogenic failure 19, 617592
CFAP44	100%	100%	100%	99.6%	Spermatogenic failure 20, 617593
CFAP45	100%	100%	100%	99.5%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP47	100%	99.8%	99.3%	75.1%	Spermatogenic failure, X-linked 3, 301059

CFAP54	100%	100%	100%	99.7%	
CFAP57	100%	100%	100%	99.3%	Spermatogenic failure 95, 620917
CFAP58	100%	100%	100%	99.5%	Spermatogenic failure 49, 619144
CFAP61	100%	100%	100%	99.4%	Spermatogenic failure 84, 620409
CFAP65	100%	100%	100%	99.1%	Spermatogenic failure 40, 618664
CFAP69	100%	100%	100%	99.7%	Spermatogenic failure 24, 617959
CFAP70	100%	100%	100%	99.7%	?Spermatogenic failure 41, 618670
CFAP91	100%	100%	100%	99.6%	Spermatogenic failure 51, 619177
CFTR	100%	100%	100%	99.5%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}
CT55	100%	98.8%	98.8%	75.7%	?Spermatogenic failure, X-linked, 7, 301106

CYLC1	100%	99.8%	99.4%	73%	{Spermatogenic failure, X-linked, 8, susceptibility to}, 301119
DAZ1	49.9%	49.5%	46.5%	32.5%	
DAZ2	49.7%	48.6%	46%	29%	
DAZ3	49.9%	48.7%	45.2%	28.4%	
DAZ4	49.5%	48%	45.6%	26.5%	
DCAF12L1	100%	100%	99.1%	70.4%	
DDX3Y	50%	49.8%	48.8%	22.1%	
DMC1	100%	100%	100%	99.7%	
DMRT1	100%	100%	100%	99.4%	
DNAAF11	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 10, 612518
DNAAF4	100%	100%	100%	99.8%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	99.8%	100%	97.5%	Ciliary dyskinesia, primary, 18, 614874
DNAAF6	100%	99.9%	99.1%	75.6%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100%	100%	100%	99%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH10	100%	100%	100%	99.4%	Spermatogenic failure 56, 619515

DNAH11	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100%	100%	100%	98.8%	Spermatogenic failure 39, 618643
DNAH2	99.9%	99.7%	100%	98.9%	Spermatogenic failure 45, 619094
DNAH3	100%	100%	100%	99.3%	
DNAH6	100%	100%	100%	99.8%	
DNAH7	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 50, 620356
DNAH8	100%	100%	100%	99.8%	Spermatogenic failure 46, 619095
DNAI1	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100%	100%	100%	98.9%	Ciliary dyskinesia, primary, 34, 617091
DNALI1	100%	100%	100%	98.7%	Spermatogenic failure 83, 620354
DNHD1	100%	100%	100%	99.4%	Spermatogenic failure 65, 619712
DPY19L2	100%	100%	100%	99.6%	Spermatogenic failure 9, 613958

DRC1	100%	100%	100%	99.1%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294
DZIP1	100%	100%	100%	99.7%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
E2F1	100%	99.3%	99.8%	96.1%	
EIF1AY	50%	49.9%	49.4%	21.9%	
FANCA	100%	100%	100%	99.4%	Fanconi anemia, complementation group A, 227650
FANCM	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FBXO43	100%	100%	100%	99.8%	Spermatogenic failure 64, 619696;Oocyte/zygote/embryo maturation arrest 12, 619697
FKBP6	100%	100%	100%	99.2%	Spermatogenic failure 77, 620103
FSIP2	100%	100%	100%	99.8%	Spermatogenic failure 34, 618153
GALNTL5	100%	100%	100%	99.8%	
GAS8	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 33, 616726
GCNA	100%	99.9%	99.1%	71.6%	Spermatogenic failure, X-linked, 4, 301077
GGN	100%	100%	100%	98.9%	Spermatogenic failure 69, 619826

GPAT2	99.5%	99.5%	99.6%	97.6%	
HENMT1	92.1%	92.1%	100%	99.9%	
HFM1	100%	100%	100%	99.7%	Premature ovarian failure 9, 615724
HSF2	100%	100%	100%	99.6%	
HSFY1	50%	50%	49.6%	28.6%	
HSFY2	50%	50%	49.5%	29.9%	
IFT74	100%	100%	100%	99.7%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
INSL3	78.8%	78.8%	100%	99.1%	Cryptorchidism, 219050
IQCN	100%	100%	100%	99.1%	Spermatogenic failure 78, 620170
IQUB	100%	100%	100%	99.8%	
KASH5	100%	100%	100%	98.8%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
KCNU1	100%	100%	100%	99.6%	Spermatogenic failure 79, 620196
KCTD19	100%	100%	100%	99%	
KDM5D	48.9%	48.7%	48.8%	20.3%	
KLHL10	100%	100%	100%	99.3%	Spermatogenic failure 11, 615081
LRRC23	100%	100%	100%	98.9%	Spermatogenic failure 92, 620848

M1AP	100%	100%	100%	99.6%	Spermatogenic failure 48, 619108
MCM8	94.4%	94.4%	100%	99.8%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	99.5%	Ovarian dysgenesis 4, 616185
MCMDC2	100%	100%	100%	99.6%	
MEI1	100%	100%	100%	99.4%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100%	100%	100%	99.8%	Premature ovarian failure 23, 620686;Spermatogenic failure 22, 617706
MLH3	100%	100%	99.9%	98.9%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MNS1	100%	100%	100%	99.7%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MOV10L1	100%	100%	100%	99.2%	?Spermatogenic failure 73, 619878
MSH4	100%	100%	100%	99.7%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MSH5	100%	100%	100%	98.8%	?Premature ovarian failure 13, 617442;Spermatogenic failure 74, 619937

NR0B1	100%	99.6%	98.7%	71.8%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NR5A1	100%	99.9%	100%	98.9%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
PDHA2	100%	100%	100%	99.6%	Spermatogenic failure 70, 619828
PICK1	100%	100%	100%	99.2%	
PIWIL2	100%	100%	100%	99.5%	
PLCZ1	100%	100%	100%	98.4%	Spermatogenic failure 17, 617214
PLD6	100%	100%	100%	99.1%	
PLK4	100%	100%	100%	99.5%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PMFBP1	100%	100%	100%	99.6%	Spermatogenic failure 31, 618112
PNLDC1	100%	100%	100%	99.5%	Spermatogenic failure 57, 619528
PRY	50%	50%	48.3%	25.5%	
PRY2	50%	50%	49.4%	28.9%	

QRICH2	100%	100%	100%	99.1%	Spermatogenic failure 35, 618341
RAD21L1	100%	100%	100%	99.7%	
RBBP7	100%	99.1%	98.5%	70.6%	
RBMY1A1	50%	50%	49.8%	46.6%	
RBMY1B	50%	49.6%	49%	40%	
RBMY1D	49.5%	48.2%	46.9%	41%	
RBMY1E	49.8%	49.2%	48.4%	42.5%	
RBMY1F	49.2%	48.6%	48.6%	31.5%	
RBMY1J	50%	49.9%	49.7%	34.2%	
RNF212	100%	100%	100%	99%	?Spermatogenic failure 62, 619673;Recombination rate QTL 1, 612042
RPS4Y2	50%	50%	49.3%	24.3%	
RSPH3	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 32, 616481
RSPH9	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 12, 612650
RXFP2	100%	100%	100%	99.9%	
SEPTIN12	100%	100%	100%	98.9%	Spermatogenic failure 10, 614822
SEPTIN4	100%	100%	100%	98.6%	
SHOC1	100%	100%	100%	99.8%	Spermatogenic failure 75, 619949
SLC26A8	100%	100%	100%	98.5%	Spermatogenic failure 3, 606766

SPACA1	100%	100%	100%	99.9%	?Spermatogenic failure 85, 620490
SPAG6	100%	100%	100%	99.7%	
SPATA16	100%	100%	100%	99.9%	?Spermatogenic failure 6, 102530
SPATA22	100%	100%	100%	99.9%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPEF2	100%	100%	100%	99.6%	Spermatogenic failure 43, 618751
SPINK2	100%	100%	100%	99.3%	?Spermatogenic failure 29, 618091
SPO11	100%	100%	100%	99.9%	
SRY	50%	50%	48.4%	22.2%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
SSX1	100%	100%	98.7%	73.4%	Spermatogenic failure, X-linked, 5, 301099
STAG3	100%	100%	100%	99.1%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STRA8	100%	100%	100%	99%	
STX2	100%	100%	100%	99.9%	
SUN5	100%	100%	100%	99.3%	Spermatogenic failure 16, 617187
SYCE1	100%	100%	100%	99.1%	?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947

SYCP2	100%	100%	100%	99.8%	Spermatogenic failure 1, 258150
SYCP3	100%	100%	100%	99.4%	Pregnancy loss, recurrent, 4, 270960;Spermatogenic failure 4, 270960
TAF4B	100%	100%	100%	99.3%	?Spermatogenic failure 13, 615841
TDRD12	100%	100%	100%	99.6%	
TDRD9	100%	100%	100%	99.1%	?Spermatogenic failure 30, 618110
TEKT3	100%	100%	100%	99.3%	Spermatogenic failure 81, 620277
TENT5D	100%	100%	99.2%	78.1%	
TERB1	100%	100%	100%	99.5%	Spermatogenic failure 60, 619646
TERB2	100%	100%	100%	99.9%	?Spermatogenic failure 59, 619645
TEX11	97.1%	96.9%	99.3%	75.3%	Spermatogenic failure, X-linked 2, 309120
TEX14	100%	100%	100%	99.7%	Spermatogenic failure 23, 617707
TEX15	100%	100%	100%	99.8%	Spermatogenic failure 25, 617960
TKTL1	100%	98.1%	99.3%	70.8%	
TRIM71	100%	100%	100%	96.4%	Hydrocephalus, congenital, 4, 618667
TSGA10	100%	100%	100%	99.7%	?Spermatogenic failure 26, 617961

TTC12	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 45, 618801
TTC21A	100%	100%	100%	99.5%	Spermatogenic failure 37, 618429
TTC29	100%	100%	100%	100%	Spermatogenic failure 42, 618745
UBR2	100%	99.7%	99.7%	99%	
USP26	100%	100%	99.6%	74.8%	Spermatogenic failure, X-linked, 6, 301101
USP9Y	50%	49.6%	49.4%	25.7%	Spermatogenic failure, Y-linked, 2, 415000
XKRY					
XKRYP7					
XRCC2	100%	100%	100%	99.5%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
ZFX	100%	99.9%	99.3%	74.1%	Intellectual developmental disorder, X-linked syndromic 37, 301118
ZMYND15	100%	100%	100%	98.7%	?Spermatogenic failure 14, 615842
ZSWIM7	88.9%	88.9%	100%	99.9%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834

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.
TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.
TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.

srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : March 17th, 2023.
This list is accurate for panel version DG 4.1.0

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors