MALE INFERTILITY PANEL DG-3.9.0 (167 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACTL7A	100.0%	100.0%	100.0%	99.7%	Spermatogenic failure 86, 620499
ACTL9	100.0%	100.0%	100.0%	99.8%	Spermatogenic failure 53, 619258
ACTRT1	100.0%	100.0%	97.3%	66.9%	
ADAD2	100.0%	100.0%	100.0%	99.5%	
ADCY10	100.0%	100.0%	100.0%	98.9%	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADGB	100.0%	99.9%	100.0%	98.2%	
ADGRG2	100.0%	99.8%	97.4%	70.1%	Congenital bilateral absence of vas deferens, X-linked, 300985
AK7	100.0%	100.0%	100.0%	97.4%	?Spermatogenic failure 27, 617965
AK9	100.0%	100.0%	100.0%	97.1%	Spermatogenic failure 89, 620705
AKAP3	100.0%	100.0%	100.0%	99.0%	Spermatogenic failure 82, 620353

AR	99.5%	99.0%	95.4%	64.4%	Androgen insensitivity, partial, with or without breast cancer, 312300;Spinal and bulbar muscular atrophy, X-linked 1, 313200;{Prostate cancer, susceptibility to}, 176807;Androgen insensitivity, 300068;Hypospadias 1, X-linked, 300633
ARMC2	100.0%	100.0%	100.0%	96.9%	Spermatogenic failure 38, 618433
AURKC	100.0%	100.0%	100.0%	98.9%	Spermatogenic failure 5, 243060
BNC1	100.0%	99.9%	100.0%	98.4%	?Premature ovarian failure 16, 618723
BPY2	50.0%	49.1%	47.3%	23.8%	
BPY2B	50.0%	48.9%	48.7%	25.1%	
BPY2C	50.0%	49.9%	48.0%	22.5%	
C14orf39	100.0%	100.0%	100.0%	96.6%	Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203
CATIP	100.0%	100.0%	100.0%	97.8%	?Spermatogenic failure 54, 619379
CATSPER1	100.0%	100.0%	100.0%	97.6%	Spermatogenic failure 7, 612997
CATSPER2	100.0%	100.0%	100.0%	99.1%	
CCDC146	100.0%	100.0%	100.0%	97.2%	

CCDC155	100.0%	100.0%	100.0%	98.8%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
CCDC34	100.0%	100.0%	100.0%	96.0%	Spermatogenic failure 76, 620084
CCDC39	100.0%	100.0%	100.0%	96.4%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100.0%	100.0%	100.0%	99.0%	Ciliary dyskinesia, primary, 15, 613808
CDC14A	100.0%	100.0%	99.9%	96.8%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDY1	50.0%	50.0%	48.5%	23.7%	
CDY1B	50.0%	49.9%	48.9%	21.0%	
CDY2A	50.0%	50.0%	48.7%	23.9%	
CDY2B	50.0%	50.0%	47.7%	19.9%	
CFAP43	100.0%	100.0%	100.0%	96.9%	Hydrocephalus, normal pressure, 1, 236690;Spermatogenic failure 19, 617592
CFAP44	100.0%	100.0%	100.0%	97.6%	Spermatogenic failure 20, 617593
CFAP47	99.8%	99.0%	97.4%	70.2%	Spermatogenic failure, X-linked 3, 301059
CFAP54	100.0%	100.0%	100.0%	97.2%	
CFAP58	100.0%	100.0%	100.0%	97.2%	Spermatogenic failure 49, 619144
CFAP61	100.0%	100.0%	100.0%	98.9%	Spermatogenic failure 84, 620409

CFAP65	100.0%	100.0%	100.0%	98.1%	Spermatogenic failure 40, 618664
CFAP69	100.0%	100.0%	100.0%	97.6%	Spermatogenic failure 24, 617959
CFAP70	100.0%	100.0%	100.0%	99.0%	?Spermatogenic failure 41, 618670
CFTR	100.0%	100.0%	100.0%	98.6%	Cystic fibrosis, 219700;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;Sweat chloride elevation without CF, ;{Hypertrypsinemia, neonatal},
CT55	100.0%	99.6%	96.8%	70.3%	?Spermatogenic failure, X-linked, 7, 301106
CYLC1	100.0%	100.0%	94.1%	57.2%	
DAZ1	50.0%	49.7%	46.9%	20.5%	
DAZ2	50.0%	49.8%	44.5%	17.2%	
DAZ3	49.9%	49.0%	42.4%	18.4%	
DAZ4	49.7%	49.0%	42.4%	15.0%	
DCAF12L1	100.0%	100.0%	99.5%	79.1%	
DDX3Y	50.0%	50.0%	48.4%	21.0%	
DMC1	100.0%	100.0%	100.0%	97.6%	
DMRT1	100.0%	100.0%	100.0%	99.3%	

DNAAF2	100.0%	100.0%	100.0%	96.4%	Ciliary dyskinesia, primary, 10, 612518
DNAAF4	100.0%	100.0%	100.0%	96.1%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100.0%	99.9%	99.9%	96.3%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH10	100.0%	100.0%	100.0%	98.7%	Spermatogenic failure 56, 619515
DNAH17	100.0%	100.0%	100.0%	99.0%	Spermatogenic failure 39, 618643
DNAH2	100.0%	99.7%	100.0%	98.9%	Spermatogenic failure 45, 619094
DNAH3	100.0%	100.0%	100.0%	98.3%	
DNAH6	100.0%	99.9%	100.0%	98.4%	
DNAH7	100.0%	100.0%	100.0%	98.4%	Ciliary dyskinesia, primary, 50, 620356
DNAH8	100.0%	99.7%	100.0%	97.8%	Spermatogenic failure 46, 619095
DNAI1	100.0%	100.0%	100.0%	99.2%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100.0%	100.0%	100.0%	97.9%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444

DNAJB13	100.0%	100.0%	100.0%	98.3%	Ciliary dyskinesia, primary, 34, 617091
DNHD1	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 65, 619712
DPY19L2	100.0%	100.0%	99.8%	95.8%	Spermatogenic failure 9, 613958
DRC1	100.0%	100.0%	100.0%	98.8%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294
DZIP1	100.0%	100.0%	100.0%	97.1%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
E2F1	100.0%	99.8%	99.8%	92.0%	
EIF1AY	50.0%	50.0%	48.1%	18.4%	
FANCA	100.0%	100.0%	100.0%	98.4%	Fanconi anemia, complementation group A, 227650
FANCM	100.0%	100.0%	100.0%	97.3%	?Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FBXO43	100.0%	100.0%	100.0%	97.9%	Spermatogenic failure 64, 619696;Oocyte/zygote/embryo maturation arrest 12, 619697
FKBP6	100.0%	100.0%	100.0%	97.1%	Spermatogenic failure 77, 620103
FSIP2	100.0%	100.0%	100.0%	96.9%	Spermatogenic failure 34, 618153
GALNTL5	100.0%	100.0%	100.0%	99.0%	

GAS8	100.0%	100.0%	100.0%	99.5%	Ciliary dyskinesia, primary, 33, 616726
GCNA	100.0%	100.0%	98.1%	72.2%	Spermatogenic failure, X-linked, 4, 301077
GGN	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 69, 619826
HSF2	100.0%	100.0%	100.0%	98.4%	
HSFY1	49.9%	49.7%	47.4%	17.2%	
HSFY2	49.9%	49.3%	46.9%	17.8%	
IFT74	100.0%	100.0%	100.0%	97.5%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IQCN	100.0%	100.0%	100.0%	99.4%	Spermatogenic failure 78, 620170
KCNU1	99.3%	98.8%	100.0%	98.2%	Spermatogenic failure 79, 620196
KCTD19	100.0%	100.0%	100.0%	98.6%	
KDM5D	48.9%	48.8%	47.9%	21.4%	
KLHL10	100.0%	100.0%	100.0%	99.6%	Spermatogenic failure 11, 615081
LRRC6	100.0%	100.0%	100.0%	98.6%	Ciliary dyskinesia, primary, 19, 614935
M1AP	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 48, 619108
MAATS1	100.0%	100.0%	100.0%	97.6%	Spermatogenic failure 51, 619177

MCM9	100.0%	100.0%	100.0%	98.3%	Ovarian dysgenesis 4, 616185
MEI1	100.0%	100.0%	100.0%	99.0%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100.0%	100.0%	100.0%	97.5%	Premature ovarian failure 23, 620686;Spermatogenic failure 22, 617706
MLH3	100.0%	100.0%	100.0%	98.3%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MNS1	100.0%	100.0%	100.0%	97.1%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MOV10L1	100.0%	100.0%	100.0%	98.5%	?Spermatogenic failure 73, 619878
MSH4	100.0%	100.0%	100.0%	98.3%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MSH5	100.0%	100.0%	100.0%	98.9%	?Premature ovarian failure 13, 617442;Spermatogenic failure 74, 619937
NR0B1	100.0%	99.8%	98.5%	73.1%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018

NR5A1	100.0%	100.0%	100.0%	98.6%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
PDHA2	100.0%	100.0%	100.0%	99.6%	Spermatogenic failure 70, 619828
PICK1	100.0%	100.0%	100.0%	99.1%	
PIH1D3	100.0%	100.0%	97.7%	67.9%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIWIL2	100.0%	100.0%	100.0%	98.9%	
PLCZ1	100.0%	100.0%	100.0%	97.2%	Spermatogenic failure 17, 617214
PLK4	100.0%	100.0%	100.0%	98.3%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PMFBP1	100.0%	100.0%	100.0%	98.3%	Spermatogenic failure 31, 618112
PNLDC1	100.0%	100.0%	100.0%	98.4%	Spermatogenic failure 57, 619528
PRY	50.0%	50.0%	47.2%	17.9%	
PRY2	50.0%	50.0%	47.9%	21.0%	
QRICH2	100.0%	100.0%	100.0%	99.4%	Spermatogenic failure 35, 618341
RAD21L1	100.0%	100.0%	100.0%	97.3%	

RBBP7	100.0%	99.7%	97.4%	70.2%	
RBMY1A1	50.0%	50.0%	49.8%	45.1%	
RBMY1B	50.0%	49.9%	47.9%	38.8%	
RBMY1D	49.5%	48.5%	47.0%	37.9%	
RBMY1E	50.0%	49.7%	48.6%	41.0%	
RBMY1F	49.3%	48.7%	47.3%	31.1%	
RBMY1J	49.6%	49.5%	48.5%	31.7%	
RNF212	100.0%	100.0%	100.0%	98.8%	?Spermatogenic failure 62, 619673;Recombination rate QTL 1, 612042
RPS4Y2	50.0%	50.0%	48.6%	19.3%	
RSPH3	100.0%	100.0%	100.0%	98.3%	Ciliary dyskinesia, primary, 32, 616481
RSPH9	100.0%	100.0%	100.0%	98.7%	Ciliary dyskinesia, primary, 12, 612650
SEPTIN12	100.0%	100.0%	100.0%	99.4%	Spermatogenic failure 10, 614822
SEPTIN4	100.0%	100.0%	100.0%	98.7%	
SHOC1	100.0%	100.0%	100.0%	97.4%	Spermatogenic failure 75, 619949
SPACA1	100.0%	100.0%	100.0%	98.7%	?Spermatogenic failure 85, 620490
SPAG6	100.0%	100.0%	100.0%	99.0%	
SPATA16	100.0%	100.0%	100.0%	98.9%	?Spermatogenic failure 6, 102530
SPATA22	100.0%	100.0%	100.0%	98.0%	

SPEF2	100.0%	100.0%	100.0%	98.3%	Spermatogenic failure 43, 618751
SPINK2	96.8%	96.8%	100.0%	97.1%	?Spermatogenic failure 29, 618091
SPO11	100.0%	100.0%	100.0%	97.2%	
SRY	50.0%	50.0%	47.1%	20.2%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
SSX1	100.0%	99.7%	96.7%	66.3%	Spermatogenic failure, X-linked, 5, 301099
STAG3	100.0%	100.0%	100.0%	98.5%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
SUN5	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 16, 617187
SYCE1	100.0%	100.0%	100.0%	99.3%	?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947
SYCP2	100.0%	100.0%	100.0%	96.3%	Spermatogenic failure 1, 258150
SYCP3	100.0%	100.0%	100.0%	97.6%	Pregnancy loss, recurrent, 4, 270960;Spermatogenic failure 4, 270960
TAF4B	100.0%	100.0%	100.0%	97.7%	?Spermatogenic failure 13, 615841
TDRD9	100.0%	100.0%	100.0%	98.5%	?Spermatogenic failure 30, 618110
TEKT3	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 81, 620277

TENT5D	100.0%	100.0%	98.4%	71.0%	
TERB1	100.0%	100.0%	100.0%	97.3%	Spermatogenic failure 60, 619646
TERB2	100.0%	100.0%	100.0%	96.5%	?Spermatogenic failure 59, 619645
TEX11	97.1%	96.8%	97.4%	69.1%	Spermatogenic failure, X-linked 2, 309120
TEX14	100.0%	100.0%	100.0%	98.5%	Spermatogenic failure 23, 617707
TEX15	100.0%	100.0%	100.0%	97.6%	Spermatogenic failure 25, 617960
TKTL1	100.0%	99.8%	98.1%	72.7%	
TRIM71	100.0%	100.0%	99.9%	97.5%	Hydrocephalus, congenital, 4, 618667
TSGA10	100.0%	100.0%	100.0%	97.9%	?Spermatogenic failure 26, 617961
TTC12	100.0%	100.0%	100.0%	99.3%	Ciliary dyskinesia, primary, 45, 618801
TTC21A	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 37, 618429
TTC29	99.6%	99.2%	100.0%	98.0%	Spermatogenic failure 42, 618745
UBR2	100.0%	99.9%	100.0%	98.2%	
USP26	100.0%	100.0%	96.1%	63.3%	Spermatogenic failure, X-linked, 6, 301101
USP9Y	49.9%	49.6%	47.7%	21.6%	Spermatogenic failure, Y-linked, 2, 415000

WDR66	100.0%	100.0%	100.0%	98.1%	Spermatogenic failure 33, 618152
XKRY					
XKRY2					
XRCC2	100.0%	100.0%	100.0%	99.1%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
ZFX	100.0%	100.0%	98.2%	73.3%	Intellectual developmental disorder, X-linked syndromic 37, 301118
ZMYND15	100.0%	100.0%	100.0%	99.0%	?Spermatogenic failure 14, 615842
ZSWIM7	90.6%	88.9%	100.0%	98.5%	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 3.9.0

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