WES MALE INFERTILITY DG 3.7

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
ACTL9	100.0%	100.0%	100.0%	99.9%	Spermatogenic failure 53, 619258
ADAD2	100.0%	100.0%	100.0%	99.8%	
ADCY10	100.0%	100.0%	100.0%	99.5%	
ADGRG2	100.0%	99.8%	98.4%	70.8%	Congenital bilateral absence of vas deferens, X-linked, 300985
AR	99.5%	99.0%	97.5%	68.7%	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	100.0%	100.0%	100.0%	98.1%	Spermatogenic failure 38, 618433
AURKC	100.0%	100.0%	100.0%	99.6%	Spermatogenic failure 5, 243060
BPY2	50.0%	49.1%	49.4%	28.8%	
BPY2B	50.0%	48.9%	47.9%	25.3%	
BPY2C	50.0%	49.9%	49.1%	27.0%	
C14orf39	100.0%	100.0%	100.0%	98.3%	Spermatogenic failure 52, 619202 ?Premature ovarian failure 18, 619203
CATIP	100.0%	100.0%	100.0%	99.4%	?Spermatogenic failure 54, 619379
CATSPER1	100.0%	100.0%	100.0%	98.4%	Spermatogenic failure 7, 612997
CATSPER2	100.0%	100.0%	100.0%	97.7%	

CCDC155	100.0%	100.0%	100.0%	99.5%	
CCDC39	100.0%	100.0%	100.0%	98.4%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100.0%	100.0%	100.0%	99.3%	Ciliary dyskinesia, primary, 15, 613808
CDC14A	100.0%	100.0%	100.0%	98.6%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDY1	50.0%	50.0%	49.4%	25.7%	
CDY1B	50.0%	49.9%	48.4%	19.2%	
CDY2A	50.0%	50.0%	48.1%	25.9%	
CDY2B	50.0%	50.0%	47.7%	19.2%	
CFAP43	100.0%	100.0%	100.0%	99.0%	Hydrocephalus, normal pressure, 1, 236690 Spermatogenic failure 19, 617592
CFAP44	100.0%	100.0%	100.0%	98.9%	Spermatogenic failure 20, 617593
CFAP58	100.0%	100.0%	100.0%	98.4%	Spermatogenic failure 49, 619144
CFAP65	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 40, 618664
CFAP69	100.0%	100.0%	100.0%	99.0%	Spermatogenic failure 24, 617959
CFTR	100.0%	100.0%	100.0%	99.4%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
DAZ1	50.0%	49.7%	48.1%	24.3%	
DAZ2	50.0%	49.8%	45.7%	19.9%	
DAZ3	49.9%	49.0%	43.2%	20.7%	
DAZ4	49.7%	49.0%	43.6%	18.0%	
DCAF12L1	100.0%	100.0%	99.3%	85.3%	
DDX3Y	50.0%	50.0%	48.9%	23.6%	
DMC1	100.0%	100.0%	100.0%	99.2%	

DMRT1	100.0%	100.0%	100.0%	99.9%	
DNAAF2	100.0%	100.0%	100.0%	99.7%	Ciliary dyskinesia, primary, 10, 612518
DNAAF4	100.0%	100.0%	100.0%	98.5%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100.0%	99.9%	100.0%	99.4%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	100.0%	100.0%	100.0%	99.7%	Spermatogenic failure 18, 617576 Ciliary dyskinesia, primary, 37, 617577
DNAH17	100.0%	100.0%	100.0%	99.6%	Spermatogenic failure 39, 618643
DNAH7	100.0%	100.0%	100.0%	99.2%	Ciliary dyskinesia, primary, 50, 620356
DNAH8	100.0%	99.7%	100.0%	99.2%	Spermatogenic failure 46, 619095
DNAI1	100.0%	100.0%	100.0%	99.5%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100.0%	100.0%	100.0%	99.2%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100.0%	100.0%	100.0%	99.4%	Ciliary dyskinesia, primary, 34, 617091
DNHD1	100.0%	100.0%	100.0%	99.7%	Spermatogenic failure 65, 619712
DPY19L2	100.0%	100.0%	100.0%	99.4%	Spermatogenic failure 9, 613958
E2F1	100.0%	99.8%	100.0%	99.5%	
EIF1AY	50.0%	50.0%	48.2%	20.4%	
FANCA	100.0%	100.0%	100.0%	99.5%	Fanconi anemia, complementation group A, 227650
FANCM	100.0%	100.0%	100.0%	98.7%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FKBP6	100.0%	100.0%	100.0%	98.2%	Spermatogenic failure 77, 620103

FSIP2	100.0%	100.0%	100.0%	98.7%	Spermatogenic failure 34, 618153
GALNTL5	100.0%	100.0%	100.0%	99.0%	
GAS8	100.0%	100.0%	100.0%	99.8%	Ciliary dyskinesia, primary, 33, 616726
GCNA	100.0%	100.0%	99.1%	74.4%	Spermatogenic failure, X-linked, 4, 301077
HSF2	100.0%	100.0%	100.0%	99.5%	
HSFY1	49.9%	49.7%	47.6%	21.0%	
HSFY2	49.9%	49.3%	47.6%	19.7%	
KDM5D	48.9%	48.8%	48.7%	24.5%	
KLHL10	100.0%	100.0%	100.0%	99.8%	Spermatogenic failure 11, 615081
LRRC6	100.0%	100.0%	100.0%	99.5%	Ciliary dyskinesia, primary, 19, 614935
M1AP	100.0%	100.0%	100.0%	99.8%	Spermatogenic failure 48, 619108
MAATS1	100.0%	100.0%	100.0%	99.1%	Spermatogenic failure 51, 619177
MCM9	100.0%	100.0%	100.0%	99.1%	Ovarian dysgenesis 4, 616185
MEI1	100.0%	100.0%	100.0%	99.6%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100.0%	100.0%	100.0%	98.6%	?Spermatogenic failure 22, 617706
MLH3	100.0%	100.0%	100.0%	99.2%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MNS1	100.0%	100.0%	100.0%	97.5%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MSH4	100.0%	100.0%	100.0%	99.3%	Premature ovarian failure 20, 619938 Spermatogenic failure 2, 108420

MSH5	100.0%	100.0%	100.0%	99.3%	?Premature ovarian failure 13, 617442 Spermatogenic failure 74, 619937
NR0B1	100.0%	99.8%	99.7%	81.5%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR5A1	100.0%	100.0%	100.0%	99.6%	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%	99.8%	Spermatogenic failure 70, 619828
PIH1D3	100.0%	100.0%	98.6%	72.7%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIWIL2	100.0%	100.0%	100.0%	99.5%	
PLCZ1	100.0%	100.0%	100.0%	98.6%	Spermatogenic failure 17, 617214
PLK4	100.0%	100.0%	100.0%	99.1%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PMFBP1	100.0%	100.0%	100.0%	99.0%	Spermatogenic failure 31, 618112
PNLDC1	100.0%	100.0%	100.0%	99.4%	Spermatogenic failure 57, 619528
PRY	50.0%	50.0%	48.2%	20.3%	
PRY2	50.0%	50.0%	47.5%	21.4%	
QRICH2	100.0%	100.0%	100.0%	99.7%	Spermatogenic failure 35, 618341
RAD21L1	100.0%	100.0%	100.0%	99.0%	
RBBP7	100.0%	99.7%	98.9%	74.1%	
RBMY1A1	50.0%	50.0%	49.3%	45.2%	
RBMY1B	50.0%	49.9%	47.9%	39.9%	

RBMY1D	49.5%	48.5%	46.1%	38.2%	
RBMY1E	50.0%	49.7%	48.2%	40.5%	
RBMY1F	49.3%	48.7%	48.3%	31.0%	
RBMY1J	49.6%	49.5%	49.2%	34.1%	
RNF212	100.0%	100.0%	100.0%	99.4%	?Spermatogenic failure 62, 619673 Recombination rate QTL 1, 612042
RPS4Y2	50.0%	50.0%	48.9%	25.4%	
RSPH3	100.0%	100.0%	100.0%	99.5%	Ciliary dyskinesia, primary, 32, 616481
RSPH9	100.0%	100.0%	100.0%	99.7%	Ciliary dyskinesia, primary, 12, 612650
SEPTIN12	100.0%	100.0%	100.0%	99.5%	Spermatogenic failure 10, 614822
SHOC1	100.0%	100.0%	100.0%	98.9%	Spermatogenic failure 75, 619949
SPAG6	100.0%	100.0%	100.0%	99.8%	
SPATA16	100.0%	100.0%	100.0%	99.5%	?Spermatogenic failure 6, 102530
SPATA22	100.0%	100.0%	100.0%	98.5%	
SPEF2	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 43, 618751
SPINK2	96.8%	96.8%	100.0%	99.9%	?Spermatogenic failure 29, 618091
SPO11	100.0%	100.0%	100.0%	99.0%	
SRY	50.0%	50.0%	49.1%	24.4%	46XY sex reversal 1, 400044
STAG3	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 61, 619672 Premature ovarian failure 8, 615723
SUN5	100.0%	100.0%	100.0%	99.6%	Spermatogenic failure 16, 617187
SYCE1	100.0%	100.0%	100.0%	99.7%	?Spermatogenic failure 15, 616950 ?Premature ovarian failure 12, 616947

SYCP2	100.0%	100.0%	100.0%	98.6%	Spermatogenic failure 1, 258150
SYCP3	100.0%	100.0%	100.0%	98.4%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
TAF4B	100.0%	100.0%	100.0%	99.4%	?Spermatogenic failure 13, 615841
TDRD9	100.0%	100.0%	100.0%	99.2%	?Spermatogenic failure 30, 618110
TERB1	100.0%	100.0%	100.0%	98.3%	Spermatogenic failure 60, 619646
TERB2	100.0%	100.0%	100.0%	99.3%	?Spermatogenic failure 59, 619645
TEX11	97.1%	96.8%	98.6%	71.9%	Spermatogenic failure, X-linked 2, 309120
TEX14	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 23, 617707
TEX15	100.0%	100.0%	100.0%	99.2%	Spermatogenic failure 25, 617960
TRIM71	100.0%	100.0%	100.0%	99.6%	Hydrocephalus, congenital, 4, 618667
TSGA10	100.0%	100.0%	100.0%	98.6%	?Spermatogenic failure 26, 617961
TTC29	99.6%	99.2%	100.0%	99.2%	Spermatogenic failure 42, 618745
UBR2	100.0%	99.9%	100.0%	99.2%	
USP26	100.0%	100.0%	96.5%	67.7%	Spermatogenic failure, X-linked, 6, 301101
USP9Y	49.9%	49.6%	48.4%	26.3%	Spermatogenic failure, Y-linked, 2, 415000
WDR66	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 33, 618152
XKRY	%	%	%	%	
XKRY2	%	%	%	%	

XRCC2	100.0%	100.0%	100.0%	99.5%	Spermatogenic failure 50, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
ZFX	100.0%	100.0%	98.7%	75.7%	
ZMYND15	100.0%	100.0%	100.0%	99.9%	?Spermatogenic failure 14, 615842
ZSWIM7	90.6%	88.9%	100.0%	99.5%	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 3.7.0.

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