MALE INFERTILITY PANEL DG-5.1.0 (217 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ACTL7A	100%	100%	100%	100%	99.9%	Spermatogenic failure 86, 620499
ACTL9	100%	100%	100%	100%	99.4%	Spermatogenic failure 53, 619258
ACTRT1	100%	100%	99.3%	93.1%	73.3%	
ACTRT3	100%	100%	99.8%	98.1%	94%	
ADAD2	100%	100%	100%	100%	99.2%	
ADCY10	100%	100%	100%	100%	99.9%	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADGB	100%	100%	100%	99.9%	99.5%	
ADGRG2	100%	100%	99.3%	91.3%	72.5%	Congenital bilateral absence of vas deferens, X-linked, 300985
AGTPBP1	96.5%	96.5%	100%	100%	99.7%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AK7	97%	96.1%	100%	100%	99.7%	?Spermatogenic failure 27, 617965
AK9	100%	100%	100%	100%	99.9%	Spermatogenic failure 89, 620705
AKAP3	100%	100%	100%	100%	99.9%	Spermatogenic failure 82, 620353
AKAP4	100%	100%	98.9%	91.3%	73.8%	

AR	100%	100%	98.4%	87.6%	66.9%	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%	100%	99.8%	Spermatogenic failure 38, 618433
ATG4D	100%	100%	100%	100%	99.2%	Spermatogenic failure 101, 621269
AURKC	100%	100%	100%	100%	99.5%	Spermatogenic failure 5, 243060
BCORL1	100%	100%	98.5%	87.1%	67.3%	Shukla-Vernon syndrome, 301029
BNC1	100%	100%	100%	100%	99.8%	?Premature ovarian failure 16, 618723
BPY2	50%	50%	45.9%	37.5%	24.4%	
BPY2B	50%	49.8%	45.9%	35.9%	21.2%	
BPY2C	50%	50%	44.7%	36.2%	24.9%	
BRWD1	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 51, 620438
C14orf39	100%	100%	100%	100%	99.7%	Spermatogenic failure 52, 619202;Premature ovarian failure 18, 619203
CALR3	100%	100%	100%	100%	99.4%	
CATIP	100%	100%	100%	100%	99.6%	?Spermatogenic failure 54, 619379
CATSPER1	100%	100%	100%	100%	98.9%	Spermatogenic failure 7, 612997
CATSPER2	100%	100%	100%	100%	99.6%	
CCDC146	100%	100%	100%	100%	99.8%	Spermatogenic failure 94, 620850

CCDC34	100%	100%	100%	100%	99.8%	Spermatogenic failure 76, 620084
CCDC39	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	100%	99.9%	99%	Ciliary dyskinesia, primary, 15, 613808
CDC14A	96.9%	96.9%	100%	100%	99.8%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDY1	50%	50%	47%	40.1%	26.1%	
CDY1B	50%	50%	47.1%	37.6%	24.5%	
CDY2A	50%	50%	49.4%	42.8%	26.8%	
CDY2B	50%	50%	48.9%	43.5%	27.5%	
CFAP221	100%	100%	100%	99.9%	99.6%	Ciliary dyskinesia, primary, 55, 279000
CFAP251	100%	100%	100%	100%	99.7%	Spermatogenic failure 33, 618152
CFAP43	100%	100%	100%	100%	99.9%	Hydrocephalus, normal pressure, 1, 236690;Spermatogenic failure 19, 617592
CFAP44	100%	100%	100%	100%	99.8%	Spermatogenic failure 20, 617593
CFAP45	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP46	100%	100%	100%	100%	99.4%	
CFAP47	100%	100%	99.4%	92.8%	75.9%	Spermatogenic failure, X-linked 3, 301059
CFAP54	100%	100%	100%	100%	99.9%	Spermatogenic failure 98, 621124;Ciliary dyskinesia, primary, 54, 621125
CFAP57	100%	100%	100%	100%	99.7%	Spermatogenic failure 95, 620917
CFAP58	100%	100%	100%	100%	99.7%	Spermatogenic failure 49, 619144

CFAP61	100%	100%	100%	100%	99.7%	Spermatogenic failure 84, 620409
CFAP65	100%	100%	100%	100%	99.2%	Spermatogenic failure 40, 618664
CFAP69	100%	100%	100%	100%	99.7%	Spermatogenic failure 24, 617959
CFAP70	100%	100%	100%	100%	99.7%	?Spermatogenic failure 41, 618670
CFAP74	100%	100%	100%	99.7%	98.6%	Ciliary dyskinesia, primary, 49, without situs inversus, 620197
CFAP91	100%	100%	100%	100%	99.7%	Spermatogenic failure 51, 619177
CFTR	100%	100%	100%	100%	99.6%	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%	100%	99.1%	92.5%	74%	?Spermatogenic failure, X-linked, 7, 301106
CYLC1	100%	100%	99.3%	91.5%	74.3%	{Spermatogenic failure, X-linked, 8, susceptibility to}, 301119
DAZ1	50%	50%	48%	39.4%	27%	
DAZ2	50%	50%	46.6%	39.6%	26.7%	
DAZ3	50%	49.9%	41.7%	31.8%	21.8%	
DAZ4	49.8%	49.5%	42.7%	32.8%	20.5%	
DCAF12L1	100%	100%	98.7%	89.7%	68.9%	
DDX3Y	50%	50%	48.5%	39.7%	21.3%	
DMC1	94.7%	94.7%	100%	100%	99.9%	

DMRT1	100%	100%	100%	100%	99.5%	
DNAAF1	96.2%	95.9%	100%	100%	99.5%	Ciliary dyskinesia, primary, 13, 613193
DNAAF11	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	100%	100%	100%	99.9%	99.2%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	100%	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	100%	100%	100%	100%	99.9%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	99.9%	100%	99.9%	99.1%	Ciliary dyskinesia, primary, 18, 614874
DNAAF6	100%	100%	99.5%	92.1%	76.8%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100%	100%	100%	100%	99.4%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH10	100%	100%	100%	100%	99.7%	Spermatogenic failure 56, 619515
DNAH11	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH12	100%	100%	100%	100%	99.7%	Spermatogenic failure 100, 621209
DNAH17	100%	100%	100%	99.9%	99.3%	Spermatogenic failure 39, 618643
DNAH2	100%	99.8%	100%	100%	99.3%	Spermatogenic failure 45, 619094
DNAH3	100%	100%	100%	100%	99.7%	
DNAH5	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH6	100%	100%	100%	100%	99.7%	

DNAH7	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 50, 620356
DNAH8	100%	100%	100%	100%	99.8%	Spermatogenic failure 46, 619095
DNAI1	95.7%	95.7%	100%	100%	99.8%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100%	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 34, 617091
DNALI1	100%	100%	100%	100%	99.4%	Spermatogenic failure 83, 620354
DNHD1	100%	100%	100%	100%	99.4%	Spermatogenic failure 65, 619712
DPY19L2	100%	100%	100%	99.8%	99%	Spermatogenic failure 9, 613958
DRC1	100%	100%	100%	100%	99.5%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294
DRC3	96.8%	96.8%	100%	99.9%	99%	
DZIP1	100%	100%	100%	100%	99.7%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
E2F1	100%	100%	100%	99.9%	99.1%	
EIF1AY	50%	50%	49.5%	40.3%	24%	
FANCA	100%	100%	100%	100%	99.5%	Fanconi anemia, complementation group A, 227650
FANCM	100%	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086

FBXO43	100%	100%	100%	100%	99.9%	Spermatogenic failure 64, 619696;Oocyte/zygote/ embryo maturation arrest 12, 619697
FKBP6	100%	100%	100%	99.9%	99.6%	Spermatogenic failure 77, 620103
FSIP2	100%	100%	100%	99.9%	99.5%	Spermatogenic failure 34, 618153
GALNTL5	100%	100%	100%	100%	99.7%	
GAS8	100%	100%	100%	99.9%	99.6%	
GCNA	100%	100%	98.8%	89.9%	71.3%	Spermatogenic failure, X-linked, 4, 301077
GGN	100%	100%	100%	99.9%	98.6%	Spermatogenic failure 69, 619826
GPAT2	99.6%	99.5%	99.6%	99.2%	97.7%	
HENMT1	92.1%	92.1%	100%	100%	99.7%	
HFM1	100%	100%	100%	100%	99.7%	Premature ovarian failure 9, 615724
HNRNPR	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073
HSF2	100%	100%	100%	100%	99.8%	
HSFY1	50%	50%	49.1%	42.7%	26.8%	
HSFY2	50%	50%	49.8%	44.4%	29%	
HYDIN	100%	99.9%	100%	99.9%	99.5%	Ciliary dyskinesia, primary, 5, 608647
IFT140	100%	100%	100%	100%	99.6%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectoder mal dysplasia 5, 621180

IFT74	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IGSF1	100%	100%	99.3%	91.1%	73.4%	Hypothyroidism, central, and testicular enlargement, 300888
INSL3	78.8%	78.8%	100%	100%	99.1%	Cryptorchidism, 219050
IQCN	100%	100%	100%	100%	99.4%	Spermatogenic failure 78, 620170
IQUB	100%	100%	100%	100%	99.6%	
ITPRID2	100%	100%	100%	100%	99.7%	
KASH5	100%	100%	100%	100%	99.5%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
KCNU1	100%	100%	100%	100%	99.8%	Spermatogenic failure 79, 620196
KCTD19	100%	100%	100%	100%	99.6%	
KDM5D	48.9%	48.9%	48.9%	39.3%	20.4%	
KLHL10	100%	100%	100%	100%	99.8%	Spermatogenic failure 11, 615081
LRRC23	100%	100%	100%	99.9%	99%	Spermatogenic failure 92, 620848
M1AP	100%	100%	100%	99.9%	99.2%	Spermatogenic failure 48, 619108
MCM8	94.4%	94.4%	100%	100%	99.8%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	100%	99.8%	Ovarian dysgenesis 4, 616185
MCMDC2	100%	100%	100%	100%	99.9%	
MEI1	100%	100%	100%	100%	99.6%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100%	100%	100%	100%	99.9%	Premature ovarian failure 23, 620686;Spermatogenic failure 22, 617706

MLH3	100%	100%	100%	99.9%	99.2%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MNS1	100%	100%	100%	99.9%	99.8%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MOV10L1	100%	100%	100%	100%	99.6%	?Spermatogenic failure 73, 619878
MSH4	100%	100%	100%	100%	99.8%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MSH5	100%	100%	100%	100%	99.3%	?Premature ovarian failure 13, 617442;Spermatogenic failure 74, 619937
NME5	86.6%	86.6%	100%	100%	100%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032
NR0B1	100%	100%	98.6%	88%	68.3%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NR5A1	98.7%	95%	100%	100%	99.5%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
ODAD3	100%	100%	100%	99.9%	98.7%	Ciliary dyskinesia, primary, 30, 616037
PDHA2	100%	100%	100%	100%	99.6%	Spermatogenic failure 70, 619828
PICK1	100%	100%	100%	100%	99.5%	

PIWIL2	98.5%	98.5%	100%	100%	99.7%	
PLCZ1	100%	100%	100%	99.7%	98.6%	Spermatogenic failure 17, 617214
PLD6	100%	100%	100%	99.8%	98.9%	
PLK4	100%	100%	100%	100%	99.9%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PMFBP1	100%	100%	100%	100%	99.6%	Spermatogenic failure 31, 618112
PNLDC1	95.2%	95.2%	100%	100%	99.6%	Spermatogenic failure 57, 619528
QRICH2	100%	100%	100%	100%	99.6%	Spermatogenic failure 35, 618341
RAD21L1	100%	100%	100%	100%	99.8%	
RBBP7	95.1%	94.2%	99.3%	90.3%	71.8%	Spermatogenic failure, X-linked, 9, 301137
RBMY1A1	50%	50%	50%	49.2%	46.1%	
RBMY1B	50%	50%	49%	45.6%	40.4%	
RBMY1D	49.8%	48.7%	47%	44.5%	39.3%	
RBMY1E	50%	50%	49.2%	47.3%	42.5%	
RBMY1F	49.7%	49.6%	48.3%	43.5%	31.3%	
RBMY1J	50%	50%	49.2%	44.7%	32.3%	
REC114	100%	100%	100%	100%	100%	Oocyte/zygote/embryo maturation arrest 10, 619176
RNF212	100%	100%	100%	99.9%	99%	?Spermatogenic failure 62, 619673;Recombination rate QTL 1, 612042
RNF212B	100%	100%	100%	99.9%	99.6%	
RPS4Y2	50%	50%	48.4%	38.8%	21.6%	
RSPH1	100%	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100%	100%	100%	99.9%	99.6%	Ciliary dyskinesia, primary, 32, 616481

RSPH4A	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 12, 612650
RXFP2	100%	100%	100%	100%	99.9%	
SEPTIN12	100%	100%	100%	100%	99%	Spermatogenic failure 10, 614822
SEPTIN4	100%	100%	100%	99.9%	99.3%	Spermatogenic failure 99, 621194
SHOC1	100%	100%	100%	100%	99.9%	Spermatogenic failure 75, 619949
SLC26A8	100%	100%	100%	100%	99.5%	Spermatogenic failure 3, 606766
SOHLH1	100%	100%	100%	100%	99.5%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX30	100%	100%	100%	100%	99.3%	
SPACA1	100%	100%	100%	100%	99.9%	?Spermatogenic failure 85, 620490
SPAG17	100%	100%	100%	100%	99.9%	?Spermatogenic failure 55, 619380
SPAG6	92.8%	92.8%	100%	100%	99.7%	
SPATA16	100%	100%	100%	100%	99.7%	?Spermatogenic failure 6, 102530
SPATA22	100%	100%	100%	100%	99.6%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPEF2	100%	100%	100%	100%	99.8%	Spermatogenic failure 43, 618751
SPEM2	100%	100%	100%	100%	99.5%	
SPINK2	100%	100%	100%	100%	99.3%	?Spermatogenic failure 29, 618091
SPO11	100%	100%	100%	100%	99.8%	
SRY	50%	50%	48.1%	37.3%	22.5%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045

SSX1	100%	100%	99.1%	90.9%	74.2%	Spermatogenic failure, X-linked, 5, 301099;Sarcoma, synovial, somatic, 300813
STAG3	100%	100%	100%	100%	99.3%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STRA8	100%	100%	100%	100%	99.7%	
STX2	100%	100%	100%	100%	99.9%	
SUN5	100%	100%	100%	100%	99.2%	Spermatogenic failure 16, 617187
SYCE1	100%	100%	100%	100%	99.3%	Premature ovarian failure 12, 616947;Spermatogenic failure 15, 616950
SYCP2	100%	100%	100%	100%	99.8%	Spermatogenic failure 1, 258150
SYCP3	100%	100%	100%	99.9%	99.6%	Pregnancy loss, recurrent, 4, 270960;Spermatogenic failure 4, 270960
TAF4B	96.2%	96.2%	100%	100%	99.6%	?Spermatogenic failure 13, 615841
TBC1D8	100%	100%	100%	99.9%	99.3%	
TDRD12	100%	100%	100%	100%	99.7%	
TDRD9	100%	100%	100%	100%	99.5%	?Spermatogenic failure 30, 618110
TEKT3	100%	100%	100%	100%	99.7%	Spermatogenic failure 81, 620277
TENT5D	100%	100%	99.7%	93.2%	75.2%	
TERB1	100%	100%	100%	100%	99.8%	Spermatogenic failure 60, 619646
TERB2	100%	100%	100%	100%	99.4%	?Spermatogenic failure 59, 619645
TEX11	97.1%	97.1%	99.5%	91.9%	73.8%	Spermatogenic failure, X-linked 2, 309120

TEX14	100%	100%	100%	100%	99.7%	Spermatogenic failure 23, 617707
TEX15	100%	100%	100%	100%	99.7%	Spermatogenic failure 25, 617960
TFDP3	100%	100%	98.6%	86.1%	66%	
TKTL1	100%	100%	99.1%	90.8%	70.6%	
TOP6BL	89.2%	89.1%	100%	100%	99.7%	Hydatidiform mole, recurrent, 4, 618432
TRIM71	100%	100%	100%	100%	98.9%	Hydrocephalus, congenital, 4, 618667
TSGA10	96.5%	96.5%	100%	100%	99.8%	?Spermatogenic failure 26, 617961
TTC12	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 45, 618801
TTC21A	100%	100%	100%	100%	99.8%	Spermatogenic failure 37, 618429
TTC29	100%	100%	100%	100%	99.9%	Spermatogenic failure 42, 618745
TTTY5						
UBR2	100%	99.9%	100%	99.8%	99.5%	
USP26	100%	100%	99%	91.8%	74.8%	Spermatogenic failure, X-linked, 6, 301101
USP9Y	50%	50%	48.9%	40.8%	23.9%	Spermatogenic failure, Y-linked, 2, 415000
XKRY						
XKRYP7						
XRCC2	100%	100%	100%	100%	99.9%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
ZFX	100%	100%	98.7%	90.4%	73.6%	Intellectual developmental disorder, X-linked syndromic 37, 301118

ZMYND15	100%	100%	100%	99.9%	99.1%	?Spermatogenic failure 14, 615842
ZSWIM7	91.9%	85.2%	100%	100%	99.6%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834

Gene symbols used follow HGNC 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 mapped against GRCh38.
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 mapped against GRCh38.
srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS 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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors