MALE INFERTILITY PANEL DG-5.2.0 (217 GENES)

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.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ACTL7A	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 86, 620499
ACTL9	100% | 100%	97% | 97%	98% | 98%	Spermatogenic failure 53, 619258
ACTRT1	99% | 99%	99% | 99%	100% | 100%	
ACTRT3	99% | 99%	94% | 94%	99% | 99%	
ADAD2	99% | 99%	94% | 94%	98% | 98%	
ADCY10	100% | 100%	98% | 97%	99% | 99%	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADGB	100% | 100%	97% | 97%	99% | 99%	
ADGRG2	99% | 99%	99% | 99%	100% | 100%	Congenital bilateral absence of vas deferens, X-linked, 300985
AGTPBP1	97% | 97%	98% | 98%	100% | 100%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AK7	96% | 96%	97% | 97%	99% | 99%	?Spermatogenic failure 27, 617965
AK9	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 89, 620705
AKAP3	99% | 99%	97% | 97%	99% | 99%	Spermatogenic failure 82, 620353
AKAP4	100% | 100%	99% | 99%	100% | 100%	
AR	97% | 97%	97% | 97%	99% | 99%	{Prostate cancer, hereditary, X-linked 3}, 301120;Androgen insensitivity, partial, with or without breast cancer, 312300;Spinal and bulbar muscular atrophy, X-linked 1, 313200;Androgen insensitivity, 300068;Hypospadias 1, X-linked, 300633
ARMC2	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 38, 618433
ATG4D	100% | 100%	96% | 96%	98% | 98%	Spermatogenic failure 101, 621269
AURKC	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 5, 243060
BCORL1	97% | 97%	98% | 98%	99% | 99%	Shukla-Vernon syndrome, 301029
BNC1	100% | 100%	97% | 97%	99% | 99%	?Premature ovarian failure 16, 618723

BPY2	0% | 0%	92% | 92%	50% | 50%	
BPY2B	7% | 3%	92% | 92%	93% | 92%	
BPY2C	14% | 12%	78% | 78%	100% | 100%	
BRWD1	100% | 100%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 51, 620438
C14orf39	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 52, 619202;Premature ovarian failure 18, 619203
CALR3	100% | 100%	97% | 97%	99% | 99%	
CATIP	100% | 100%	96% | 96%	98% | 98%	?Spermatogenic failure 54, 619379
CATSPER1	100% | 100%	94% | 94%	99% | 99%	Spermatogenic failure 7, 612997
CATSPER2	99% | 99%	97% | 97%	99% | 99%	
CCDC146	99% | 99%	96% | 96%	99% | 99%	Spermatogenic failure 94, 620850
CCDC34	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 76, 620084
CCDC39	100% | 100%	98% | 98%	99% | 99%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99% | 99%	94% | 94%	98% | 98%	Ciliary dyskinesia, primary, 15, 613808
CDC14A	97% | 97%	98% | 98%	99% | 99%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDY1	0% | 0%	100% | 100%	90% | 80%	
CDY1B	0% | 0%	100% | 100%	100% | 100%	
CDY2A	1% | 0%	85% | 85%	100% | 100%	
CDY2B	0% | 0%	92% | 92%	100% | 100%	
CFAP221	100% | 100%	96% | 96%	99% | 99%	Ciliary dyskinesia, primary, 55, 279000
CFAP251	99% | 99%	96% | 96%	99% | 99%	Spermatogenic failure 33, 618152
CFAP43	100% | 100%	98% | 98%	99% | 99%	Hydrocephalus, normal pressure, 1, 236690;Spermatogenic failure 19, 617592
CFAP44	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 20, 617593
CFAP45	100% | 100%	96% | 96%	99% | 99%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP46	100% | 100%	95% | 95%	99% | 99%	
CFAP47	100% | 100%	99% | 99%	100% | 100%	Spermatogenic failure, X-linked 3, 301059
CFAP54	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 98, 621124;Ciliary dyskinesia, primary, 54, 621125
CFAP57	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 95, 620917
CFAP58	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 49, 619144
CFAP61	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 84, 620409

CFAP65	100% | 100%	97% | 97%	98% | 98%	Spermatogenic failure 40, 618664
CFAP69	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 24, 617959
CFAP70	100% | 100%	98% | 98%	100% | 100%	?Spermatogenic failure 41, 618670
CFAP74	99% | 99%	94% | 94%	97% | 97%	Ciliary dyskinesia, primary, 49, without situs inversus, 620197
CFAP91	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 51, 619177
CFTR	100% | 100%	98% | 98%	100% | 100%	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	66% | 66%	82% | 82%	100% | 100%	?Spermatogenic failure, X-linked, 7, 301106
CYLC1	100% | 100%	99% | 99%	100% | 100%	{Spermatogenic failure, X-linked, 8, susceptibility to}, 301119
DAZ1	2% | 0%	85% | 85%	99% | 99%	
DAZ2	3% | 3%	93% | 93%	84% | 82%	
DAZ3	4% | 1%	96% | 95%	93% | 93%	
DAZ4	3% | 3%	89% | 89%	83% | 83%	
DCAF12L1	99% | 99%	98% | 98%	100% | 100%	
DDX3Y	90% | 90%	100% | 100%	100% | 100%	
DMC1	95% | 95%	97% | 97%	99% | 99%	
DMRT1	100% | 100%	96% | 96%	99% | 99%	
DNAAF1	95% | 94%	96% | 96%	98% | 98%	Ciliary dyskinesia, primary, 13, 613193
DNAAF11	100% | 100%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	100% | 100%	96% | 96%	98% | 98%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99% | 99%	94% | 94%	96% | 96%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	100% | 100%	97% | 97%	99% | 99%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	98% | 98%	95% | 95%	99% | 99%	Ciliary dyskinesia, primary, 18, 614874
DNAAF6	100% | 100%	99% | 99%	100% | 100%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH10	100% | 100%	97% | 97%	99% | 98%	Spermatogenic failure 56, 619515
DNAH11	100% | 100%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH12	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 100, 621209

DNAH17	100% | 100%	96% | 96%	96% | 95%	Spermatogenic failure 39, 618643
DNAH2	100% | 99%	96% | 96%	99% | 99%	Spermatogenic failure 45, 619094
DNAH3	100% | 100%	97% | 97%	99% | 99%	
DNAH5	100% | 100%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH6	100% | 100%	97% | 97%	99% | 99%	
DNAH7	100% | 100%	98% | 98%	100% | 100%	Ciliary dyskinesia, primary, 50, 620356
DNAH8	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 46, 619095
DNAI1	96% | 96%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100% | 100%	95% | 95%	98% | 98%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100% | 100%	96% | 96%	98% | 98%	Ciliary dyskinesia, primary, 34, 617091
DNALI1	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 83, 620354
DNHD1	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 65, 619712
DPY19L2	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 9, 613958
DRC1	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294
DRC3	95% | 95%	93% | 93%	99% | 99%	
DZIP1	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
E2F1	99% | 99%	94% | 94%	99% | 99%	
EIF1AY	90% | 90%	100% | 100%	100% | 100%	
FANCA	100% | 100%	97% | 97%	99% | 99%	Fanconi anemia, complementation group A, 227650
FANCM	100% | 100%	98% | 98%	100% | 100%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FBXO43	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 64, 619696;Oocyte/zygote/embryo maturation arrest 12, 619697
FKBP6	100% | 100%	96% | 96%	98% | 98%	Spermatogenic failure 77, 620103
FSIP2	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 34, 618153
GALNTL5	100% | 100%	98% | 98%	99% | 99%	
GAS8	100% | 100%	96% | 96%	98% | 98%	
GCNA	99% | 99%	97% | 97%	100% | 100%	Spermatogenic failure, X-linked, 4, 301077
GGN	100% | 100%	96% | 96%	98% | 98%	Spermatogenic failure 69, 619826
GPAT2	67% | 66%	74% | 71%	99% | 99%	

HENMT1	93% | 93%	98% | 98%	99% | 99%	
HFM1	100% | 100%	98% | 98%	99% | 99%	?Hydatidiform mole, recurrent, 5, 621720;Premature ovarian failure 9, 615724
HNRNPR	100% | 100%	97% | 97%	99% | 99%	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073
HSF2	100% | 100%	98% | 98%	100% | 100%	
HSFY1	0% | 0%	97% | 97%	99% | 95%	
HSFY2	1% | 1%	98% | 98%	100% | 100%	
HYDIN	88% | 85%	93% | 91%	99% | 99%	Ciliary dyskinesia, primary, 5, 608647
IFT140	100% | 100%	96% | 96%	98% | 98%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT74	100% | 100%	98% | 98%	99% | 99%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IGSF1	98% | 98%	98% | 98%	100% | 100%	Hypothyroidism, central, and testicular enlargement, 300888
INSL3	80% | 80%	95% | 94%	97% | 97%	Cryptorchidism, 219050
IQCN	100% | 100%	96% | 96%	97% | 97%	Spermatogenic failure 78, 620170
IQUB	100% | 100%	98% | 98%	99% | 99%	
ITPRID2	100% | 100%	98% | 98%	100% | 100%	
KASH5	100% | 100%	97% | 96%	98% | 98%	Hydatidiform mole, recurrent, 6, 621721;Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
KCNU1	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 79, 620196
KCTD19	100% | 100%	97% | 97%	99% | 99%	
KDM5D	87% | 86%	100% | 100%	100% | 100%	
KLHL10	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 11, 615081
LRRC23	99% | 99%	96% | 96%	99% | 99%	Spermatogenic failure 92, 620848
M1AP	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 48, 619108
MCM8	94% | 94%	98% | 98%	100% | 100%	?Premature ovarian failure 10, 612885
MCM9	100% | 100%	97% | 97%	99% | 99%	Ovarian dysgenesis 4, 616185
MCMDC2	100% | 100%	98% | 98%	100% | 100%	
MEI1	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 103, 621619;Oocyte/zygote/embryo maturation arrest 26, 621620;Hydatidiform mole, recurrent, 3, 618431
MEIOB	100% | 100%	98% | 98%	99% | 99%	Premature ovarian failure 23, 620686;Spermatogenic failure 22, 617706

MLH3	100% | 100%	97% | 97%	99% | 99%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MNS1	100% | 100%	98% | 98%	99% | 99%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MOV10L1	100% | 100%	96% | 96%	98% | 98%	?Spermatogenic failure 73, 619878
MSH4	100% | 100%	98% | 98%	99% | 99%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MSH5	100% | 100%	96% | 96%	99% | 98%	?Premature ovarian failure 13, 617442;Spermatogenic failure 74, 619937
NME5	87% | 87%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032
NR0B1	98% | 98%	97% | 97%	100% | 100%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NR5A1	94% | 93%	96% | 96%	98% | 98%	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%	95% | 95%	97% | 97%	Ciliary dyskinesia, primary, 30, 616037
PDHA2	100% | 100%	97% | 97%	100% | 100%	Spermatogenic failure 70, 619828
PICK1	100% | 100%	96% | 96%	99% | 95%	
PIWIL2	99% | 99%	98% | 98%	99% | 99%	
PLCZ1	99% | 99%	96% | 96%	99% | 99%	Spermatogenic failure 17, 617214
PLD6	100% | 100%	97% | 97%	99% | 99%	
PLK4	100% | 100%	98% | 98%	99% | 99%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PMFBP1	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 31, 618112
PNLDC1	95% | 95%	96% | 96%	99% | 99%	Spermatogenic failure 57, 619528
QRICH2	99% | 99%	95% | 95%	98% | 98%	Spermatogenic failure 35, 618341
RAD21L1	100% | 100%	98% | 98%	99% | 99%	
RBBP7	93% | 93%	99% | 99%	100% | 100%	Spermatogenic failure, X-linked, 9, 301137
RBMY1A1	34% | 33%	94% | 94%	100% | 100%	
RBMY1B	0% | 0%	100% | 100%	100% | 100%	
RBMY1D	0% | 0%	87% | 87%	99% | 99%	
RBMY1E	21% | 21%	86% | 85%	100% | 100%	
RBMY1F	12% | 9%	89% | 88%	96% | 91%	
RBMY1J	9% | 8%	81% | 81%	100% | 100%	
REC114	100% | 100%	98% | 98%	100% | 100%	Oocyte/zygote/embryo maturation arrest 10, 619176

RNF212	98% | 98%	91% | 91%	98% | 98%	?Spermatogenic failure 62, 619673;Recombination rate QTL 1, 612042
RNF212B	100% | 100%	97% | 97%	99% | 99%	
RPS4Y2	90% | 90%	100% | 100%	100% | 100%	
RSPH1	100% | 100%	93% | 92%	99% | 99%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100% | 100%	94% | 94%	99% | 99%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	100% | 100%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100% | 100%	97% | 97%	99% | 99%	Ciliary dyskinesia, primary, 12, 612650
RXFP2	100% | 100%	98% | 98%	99% | 99%	
SEPTIN12	100% | 100%	95% | 95%	98% | 98%	Spermatogenic failure 10, 614822
SEPTIN4	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 99, 621194
SHOC1	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 75, 619949
SLC26A8	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 3, 606766
SOHLH1	99% | 99%	95% | 95%	97% | 97%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX30	100% | 100%	97% | 97%	99% | 99%	
SPACA1	100% | 100%	98% | 98%	100% | 100%	?Spermatogenic failure 85, 620490
SPAG17	100% | 100%	98% | 98%	100% | 100%	?Spermatogenic failure 55, 619380
SPAG6	93% | 93%	97% | 97%	100% | 100%	
SPATA16	100% | 100%	97% | 97%	99% | 99%	?Spermatogenic failure 6, 102530
SPATA22	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPEF2	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 43, 618751
SPEM2	100% | 100%	97% | 97%	98% | 98%	
SPINK2	100% | 100%	97% | 97%	98% | 98%	?Spermatogenic failure 29, 618091
SPO11	100% | 100%	98% | 98%	99% | 99%	
SRY	89% | 89%	100% | 100%	100% | 100%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
SSX1	100% | 100%	98% | 98%	100% | 100%	Spermatogenic failure, X-linked, 5, 301099;Sarcoma, synovial, somatic, 300813
STAG3	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STRA8	99% | 99%	95% | 94%	99% | 99%	
STX2	99% | 99%	96% | 96%	99% | 99%	
SUN5	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 16, 617187

SYCE1	99% | 99%	96% | 96%	99% | 99%	Premature ovarian failure 12, 616947;Spermatogenic failure 15, 616950
SYCP2	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 1, 258150
SYCP3	100% | 100%	97% | 97%	99% | 99%	Pregnancy loss, recurrent, 4, 270960;Spermatogenic failure 4, 270960
TAF4B	96% | 96%	97% | 97%	100% | 100%	?Spermatogenic failure 13, 615841
TBC1D8	99% | 99%	96% | 96%	99% | 99%	
TDRD12	100% | 100%	97% | 97%	99% | 99%	
TDRD9	100% | 100%	97% | 97%	99% | 99%	?Spermatogenic failure 30, 618110
TEKT3	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 81, 620277
TENT5D	100% | 100%	99% | 99%	100% | 100%	
TERB1	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 60, 619646
TERB2	100% | 100%	97% | 97%	99% | 99%	?Spermatogenic failure 59, 619645
TEX11	97% | 97%	98% | 98%	100% | 100%	Spermatogenic failure, X-linked 2, 309120
TEX14	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 23, 617707
TEX15	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 25, 617960
TFDP3	100% | 100%	99% | 99%	100% | 100%	
TKTL1	98% | 98%	99% | 99%	100% | 100%	
TOP6BL	76% | 76%	97% | 97%	99% | 99%	Hydatidiform mole, recurrent, 4, 618432
TRIM71	100% | 100%	95% | 95%	99% | 99%	Hydrocephalus, congenital, 4, 618667
TSGA10	97% | 97%	97% | 97%	100% | 100%	?Spermatogenic failure 26, 617961
TTC12	100% | 100%	97% | 97%	100% | 100%	Ciliary dyskinesia, primary, 45, 618801
TTC21A	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 37, 618429
TTC29	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 42, 618745
TTTY5				
UBR2	100% | 100%	97% | 97%	99% | 99%	
USP26	100% | 100%	98% | 98%	100% | 100%	Spermatogenic failure, X-linked, 6, 301101
USP9Y	89% | 89%	100% | 100%	100% | 100%	Spermatogenic failure, Y-linked, 2, 415000
XKRY				
XKRYP7				

XRCC2	100% | 100%	97% | 97%	99% | 99%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
ZFX	100% | 100%	99% | 99%	100% | 100%	Intellectual developmental disorder, X-linked syndromic 37, 301118
ZMYND15	100% | 100%	95% | 95%	98% | 98%	?Spermatogenic failure 14, 615842
ZSWIM7	85% | 83%	97% | 97%	99% | 99%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834