HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL DG-5.1.0 (77 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*
ADCY3	100%	100%	100%	100%	99.5%	{Obesity, susceptibility to, BMIQ19}, 617885
ANOS1	100%	100%	98.7%	89.8%	71.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ARHGAP35	100%	100%	100%	100%	99.5%	
ARL6	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
AXL	100%	100%	100%	100%	99.1%	
BBS1	93.7%	93.7%	100%	100%	99.3%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	100%	99.8%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 7, 615984
CCDC141	100%	100%	100%	100%	99.8%	

CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CNGA2	100%	99.9%	98%	87.1%	68.1%	
CUL4B	96.7%	96.7%	99.2%	91.3%	74%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080
DCC	100%	100%	100%	100%	99.7%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Esophageal carcinoma, somatic, 133239;Colorectal cancer, somatic, 114500;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DUSP6	100%	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EMX2	100%	100%	100%	99.7%	96%	Schizencephaly, 269160
FEZF1	100%	100%	100%	99.9%	99.6%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100%	100%	100%	100%	99.5%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	100%	100%	100%	99.8%	98.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

FGFR1	100%	99.2%	100%	100%	99.4%	Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FLRT3	100%	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	100%	100%	100%	100%	99.6%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GLI2	100%	100%	100%	99.9%	99.3%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GNRH1	100%	100%	100%	100%	99.8%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	100%	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HS6ST1	100%	100%	100%	100%	98.6%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880

IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	99.8%	96.5%	100%	100%	99.9%	Bardet-Biedl syndrome 19, 615996
IFT74	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IGSF10	100%	100%	100%	100%	99.8%	
IL17RD	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KISS1	100%	100%	100%	100%	99.5%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	100%	100%	100%	98.8%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400
KLB	100%	100%	100%	100%	99.8%	
LEP	100%	100%	100%	100%	99.5%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94.6%	94.6%	100%	100%	99.7%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	100%	100%	100%	99.8%	98%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300

LHX3	100%	100%	100%	99.6%	98.9%	Pituitary hormone deficiency, combined, 3, 221750
LZTFL1	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 17, 615994
MKKS	100%	100%	100%	100%	100%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
NDNF	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NOS1	100%	100%	100%	100%	99.2%	
NR0B1	100%	100%	98.6%	88%	68.3%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NSMF	100%	100%	100%	100%	99.2%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NTN1	100%	100%	100%	100%	99.1%	Mirror movements 4, 618264
PCSK1	100%	100%	100%	100%	99.9%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PHF6	100%	100%	99.4%	92.6%	74.6%	Borjeson-Forssman-Lehmann syndrome, 301900
PLXNA1	100%	100%	100%	100%	99.6%	Dworschak-Punetha neurodevelopmental syndrome, 619955

PNPLA6	100%	100%	100%	100%	99.3%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
POLG	100%	100%	100%	100%	99.8%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR3A	100%	100%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742

POLR3GL	100%	100%	100%	100%	99.5%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
PROK2	100%	100%	100%	100%	100%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100%	100%	100%	99.9%	99.1%	Pituitary hormone deficiency, combined, 2, 262600
RNF216	100%	100%	100%	100%	99.7%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
SDCCAG8	100%	100%	100%	100%	99.9%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SEMA3A	100%	100%	100%	100%	99.8%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	100%	100%	100%	100%	99.9%	
SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100%	100%	100%	99.7%	96%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866

SOX2	100%	100%	100%	99.9%	97.8%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SPRY4	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100%	100%	100%	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TCF12	96.1%	96.1%	100%	100%	99.7%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TENM1	100%	100%	99.2%	91%	72.1%	
TRIM32	100%	100%	100%	100%	99.6%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TTC8	91.4%	91.4%	100%	100%	99.9%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
WDPCP	92.1%	92.1%	100%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085

WDR11	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858

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