WES HYPOGONADOTROPIC HYPOGONADISM (KALLMANN)

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*
ADCY3	100.0%	100.0%	100.0%	99.6%	
ANOS1	100.0%	99.8%	99.1%	73.9%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ARHGAP35	100.0%	100.0%	100.0%	99.4%	
AXL	100.0%	100.0%	100.0%	99.6%	
CCDC141	99.5%	98.9%	100.0%	99.1%	
CHD7	100.0%	100.0%	100.0%	99.5%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CNGA2	99.9%	99.7%	98.6%	72.1%	
DCAF17	100.0%	100.0%	100.0%	99.8%	Woodhouse-Sakati syndrome, 241080
DCC	100.0%	100.0%	100.0%	99.5%	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	100.0%	100.0%	100.0%	99.3%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269

FEZF1	100.0%	100.0%	100.0%	99.0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100.0%	100.0%	100.0%	99.9%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	100.0%	100.0%	100.0%	99.7%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100.0%	100.0%	100.0%	99.8%	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	100.0%	99.7%	100.0%	99.9%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	98.7%	98.0%	100.0%	100.0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GNRH1	100.0%	100.0%	100.0%	97.9%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	100.0%	100.0%	100.0%	97.4%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HS6ST1	100.0%	100.0%	100.0%	99.5%	
IGSF10	100.0%	100.0%	100.0%	99.6%	

IL17RD	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KISS1	100.0%	100.0%	100.0%	95.9%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100.0%	100.0%	100.0%	99.9%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KLB	100.0%	100.0%	100.0%	99.7%	
LEP	100.0%	100.0%	100.0%	99.4%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94.6%	94.6%	100.0%	99.3%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	100.0%	100.0%	100.0%	100.0%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHX3	100.0%	100.0%	100.0%	99.8%	Pituitary hormone deficiency, combined, 3, 221750
NDNF	100.0%	100.0%	100.0%	99.0%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NOS1	100.0%	100.0%	100.0%	99.6%	
NR0B1	100.0%	99.8%	99.7%	81.5%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NSMF	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NTN1	100.0%	100.0%	100.0%	99.6%	Mirror movements 4, 618264
PCSK1	100.0%	100.0%	100.0%	99.5%	Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PLXNA1	100.0%	100.0%	100.0%	100.0%	Dworschak-Punetha neurodevelopmental syndrome, 619955

POLG	100.0%	100.0%	100.0%	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
PROK2	100.0%	100.0%	100.0%	99.7%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100.0%	100.0%	99.9%	96.3%	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	100.0%	100.0%	100.0%	99.7%	
SEMA3E	100.0%	100.0%	100.0%	99.7%	
SOX10	100.0%	100.0%	100.0%	99.9%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX2	100.0%	100.0%	100.0%	99.4%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SPRY4	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	100.0%	100.0%	100.0%	99.9%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839

TACR3	100.0%	99.8%	100.0%	99.3%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TCF12	100.0%	100.0%	100.0%	99.6%	Craniosynostosis 3, 615314 Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TENM1	99.9%	99.5%	98.8%	75.0%	
WDR11	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal recessive 78, 620237 Hypogonadotropic hypogonadism 14 with or without anosmia, 614858

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