HYPOGONADOTROPIC HYPOGONADISM DG 2.16 (37 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ADCY3	117,3	99.9%	98.7%	{Obesity, susceptibility to, BMIQ19}, 617885
ANOS1	76,7	91.7%	88.0%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
CCDC141	111,9	99.9%	98.6%	No OMIM phenotype
CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
DCC	118,6	100.0%	99.8%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DUSP6	164,1	100.0%	100.0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FEZF1	179,7	100.0%	100.0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	167,3	100.0%	100.0%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	130	97.9%	86.8%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	122,6	100.0%	99.6%	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FLRT3	170,3	100.0%	100.0%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	112	100.0%	100.0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GNRH1	84,5	96.5%	78.0%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	145,4	100.0%	100.0%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	66,2	100.0%	98.7%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HS6ST1	72,8	97.7%	92.0%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880

IGSF10	186,5	100.0%	99.9%	No OMIM phenotype
IL17RD	134	99.9%	99.0%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KISS1	79,5	100.0%	98.1%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	156,4	100.0%	100.0%	?Precocious puberty, central, 1, 176400 Hypogonadotropic hypogonadism 8 with or without anosmia, 614837
KLB	211,9	100.0%	100.0%	No OMIM phenotype
LEP	174,7	100.0%	99.9%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	106,5	94.2%	91.4%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	23,5	92.3%	52.6%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
NR0B1	138,6	99.9%	99.2%	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, 300200
NSMF	109,8	99.4%	97.4%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	141,9	100.0%	99.2%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PROK2	117,4	100.0%	99.9%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	223	100.0%	100.0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	96,9	92.5%	83.7%	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	157,2	100.0%	100.0%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SOX10	88,2	100.0%	99.1%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPRY4	164,5	100.0%	99.6%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	61,8	99.9%	95.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	146,1	100.0%	99.7%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TCF12	137,7	99.9%	99.9%	Craniosynostosis 3, 615314
WDR11	115	97.5%	96.2%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors