HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 2.11 (33 genes)

Gene	*Median*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ADCY3	129.7	99	98	No OMIM phenotype
CCDC141	123.3	99	98	No OMIM phenotype
CHD7	150.9	99	98	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
DUSP6	176.2	100	99	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FEZF1	159	99	99	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	139	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	111.5	90	79	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	148.3	99	98	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	225.8	100	100	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	148.8	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GNRH1	86.6	99	91	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	162	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	57.8	99	92	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HS6ST1	75.5	94	85	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
IL17RD	133.9	99	97	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KAL1	90.9	89	87	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KISS1	42.7	99	95	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842

KISS1R	106.6	99	95	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
LEP	188.9	100	99	Obesity, morbid, due to leptin deficiency, 614962
LEPR	109.7	93	90	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	29	97	73	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
NR0B1	120.1	99	98	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200
NSMF	94.8	96	95	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	147.1	100	99	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PROK2	105.6	98	91	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	332	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	76.6	91	84	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	182	100	100	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SOX10	65.8	98	91	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPRY4	139.1	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	80.5	99	90	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	180.5	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
WDR11	130.7	96	96	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 : April 14th, 2017.
This list is accurate for panel version DG 2.11
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors