

HYPOGONADOTROPIC HYPOGONADISM DG 2.15 (37 genes)

Releasedate: 31-01-2019

<i>Gene</i>	<i>Median Coverage</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
ADCY3	130.4	99.8	98.4	{Obesity, susceptibility to, BMIQ19}, 617885
ANOS1	90.3	89.4	87.6	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
CCDC141	111.9	99.9	98.0	No OMIM phenotype PMID:27014940,PMID: 28324054
CHD7	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
DCC	138.5	100.0	99.9	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	175.9	100.0	99.9	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FEZF1	158.8	99.9	99.3	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	138.6	100.0	100.0	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	111.4	90.2	79.7	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

FGFR1	148	99.7	98.3	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	225.7	100.0	100.0	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	149.1	100.0	100.0	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GNRH1	86.7	99.5	91.5	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	161.9	100.0	100.0	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	57.6	99.2	92.6	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HS6ST1	75.3	94.8	85.0	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
IGSF10	246.8	100.0	99.9	No OMIM phenotype Delayed puberty, Howard et al. EMBO Mol Med. 2016 Jun 1 8(6):626-42
IL17RD	135.6	99.3	97.7	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KISS1	41.4	98.5	91.2	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	106.4	99.5	95.3	?Precocious puberty, central, 1, 176400 Hypogonadotropic hypogonadism 8 with or without anosmia, 614837

KLB	213.4	100.0	100.0	No OMIM phenotype Xu et al. EMBO Mol Med. 2017 Oct 9(10):1379-1397
LEP	188.8	100.0	99.6	Obesity, morbid, due to leptin deficiency, 614962
LEPR	109.6	93.8	90.2	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	29	97.2	73.2	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
NROB1	119.3	99.9	98.6	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, 300200
NSMF	78.4	95.7	95.2	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	147.2	100.0	99.0	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PROK2	105.6	98.4	91.9	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	331.8	100.0	100.0	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	76.5	91.6	84.3	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	182	100.0	100.0	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SOX10	65.8	98.2	91.3	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPRY4	138.7	100.0	100.0	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266

TAC3	80.5	99.1	91.1	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	180.3	100.0	100.0	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TCF12	150.3	100.0	99.8	Craniosynostosis 3, 615314
WDR11	130.6	96.9	96.4	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.

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 : December 31st, 2018.

This list is accurate for panel version DG 2.15

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors
