HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
CHD7	161	99%	98%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
DUSP6	173.3	100%	99%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FEZF1	171.3	99%	99%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	155.4	99%	99%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	127.4	85%	79%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	161.5	99%	97%	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	259.8	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	150	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GNRH1	102.6	99%	92%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	210.9	100%	100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	78.1	96%	87%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230
HS6ST1	68.9	93%	84%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
IL17RD	144.8	99%	97%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KAL1	106.4	89%	87%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KISS1	47.2	98%	94%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	93.7	99%	97%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
LEP	214	100%	98%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	125.5	93%	89%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	28.3	94%	68%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300

NR0B1	130.1	99%	97%	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200
NSMF	113	96%	95%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	166.9	99%	99%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PROK2	106.7	97%	91%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	373.6	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	98.3	97%	91%	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	214.9	100%	99%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SOX10	80.4	97%	93%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPRY4	153	99%	99%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	88.5	96%	88%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	179.2	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
WDR11	148.7	97%	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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