

# HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 2.9

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
CHD7	168.4	100%	99%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
DUSP6	172.7	100%	99%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FEZF1	170.4	100%	99%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	167.7	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	137.4	87%	80%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	165.2	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	242.5	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	158.9	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GNRH1	100.5	98%	89%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	218.3	100%	100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	96.7	99%	96%	Growth hormone deficiency with pituitary anomalies, 182230 Pituitary hormone deficiency, combined, 5, 182230 Septo-optic dysplasia, 182230
HS6ST1	71.3	94%	83%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
IL17RD	146.4	99%	98%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KAL1	102.2	89%	87%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KISS1	51.5	100%	97%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	97.2	99%	98%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
LEP	228.8	100%	99%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	138.5	94%	92%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	26.1	95%	64%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300

NROB1	130	99%	98%	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200
NSMF	116.6	96%	95%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	175.4	100%	99%	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362
PROK2	116.6	99%	92%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	387.8	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	92.2	94%	86%	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	231	100%	100%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SOX10	74.6	98%	95%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPRY4	146.5	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	80.1	98%	90%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	178.6	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
WDR11	142.6	97%	96%	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 : April 14<sup>th</sup> 2017

This list is accurate for panel version DG 2.9

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