

# HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 2.3.x

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
CHD7	115,7	100%	98%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 {Scoliosis, idiopathic 3}, 608765
DUSP6	146,1	100%	99%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FGF17	99,2	100%	97%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	47,2	85%	67%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	111,5	100%	95%	Hartsfield syndromic, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FLRT3	195,3	100%	100%	Hypogonadotropic hypogonadism 21 with or without anosmia, 615271
FSHB	93,5	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia
GNRH1	38,4	95%	77%	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	156,3	100%	100%	Hypogonadotropic hypogonadism 7 with or without anosmia, 138850
HS6ST1	6,9	23%	0%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
IL17RD	99,2	98%	96%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KAL1	44,3	88%	78%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KISS1	43,5	96%	88%	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	42,3	100%	90%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
NROB1	56,9	100%	100%	Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NSMF	103,9	96%	95%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	105,6	100%	93%	Obesity with impaired prohormone processing, 60955 {Obesity, susceptibility to, BMIQ12}, 612362

PROK2	95,2	100%	97%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	168,4	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	64,8	100%	83%	Pituitary hormone deficiency, combined, 2
SEMA3A	139,7	100%	100%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SOX10	65,1	100%	97%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurological involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPRY4	89	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	76,9	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	149	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
WDR11	86,6	99%	98%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858

*Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014*

*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*

*OMIM release used for OMIM disease identifiers and descriptions : June 30<sup>th</sup>, 2015*

*This list is accurate for all panel versions starting with DG 2.3. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)*

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

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