

# HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 2.5/2.6

<i>Gene name</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated phenotype description and OMIM disease ID</i>
ADCY3	126.2	99%	98%	No OMIM phenotype
CHD7	144.8	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	168.5	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FGF17	143.6	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	107.9	82%	76%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	148.8	100%	97%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465
FLRT3	245.2	100%	100%	Hypogonadotropic hypogonadism 21 with or without anosmia, 615271
FSHB	147.8	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia,229070
GNRH1	101.1	99%	88%	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	206.1	100%	100%	Hypogonadotropic hypogonadism 7 with or without anosmia, 138850
HS6ST1	65.8	96%	85%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
IL17RD	140	100%	99%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KAL1	133	89%	87%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KISS1	40.4	99%	99%	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	93.9	100%	96%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty,central,1,176400
NROB1	140	100%	99%	Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism,300200 46XY sex reversal 2,dosage-sensitive,300018
NSMF	109.6	98%	96%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838

PCSK1	147.1	100%	99%	Obesity with impaired prohormone processing,60955 {Obesity,susceptibility to,BMIQ12},612362
PROK2	88	99%	99%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	363.4	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	81.9	96%	93%	Pituitary hormone deficiency, combined, 2,262600
SEMA3A	196.1	100%	100%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SOX10	69.4	100%	89%	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	139	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	80.3	99%	91%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	175.2	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
WDR11	137.3	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 10th, 2016.*

*This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.*

*Ad 1. "No OMIM phenotype" signifies a gene without*