

# HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DGD141114

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM ID</i>
CHD7	117.2	100%	99%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 {Scoliosis, idiopathic 3}, 608765
DUSP6	147.5	100%	98%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FGF17	95.9	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	52.7	84%	68%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	115.2	99%	96%	Hypogonadotropic hypogonadism 2 with or without anosmia, 147950
FLRT3	195.3	100%	100%	Hypogonadotropic hypogonadism 21 with or without anosmia, 615271
GNRH1	40.4	100%	97%	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	157.4	100%	100%	Hypogonadotropic hypogonadism 7 with or without anosmia, 138850
HS6ST1	6.3	24%	%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
IL17RD	101.4	100%	97%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KAL1	76.6	96%	91%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KISS1	35.5	88%	74%	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	43.7	98%	87%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837
NSMF	86.1	96%	95%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PROK2	95.4	100%	91%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	174.5	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
SEMA3A	138.8	100%	100%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897

SOX10	67.6	100%	100%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurological involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPRY4	93.8	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	77.0	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	154.7	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
WDR11	92.3	100%	99%	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 : 31 october 2014*

*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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