# HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 2.17 (45 genes)

Releasedate: 06-12-2019

| *Gene* | *Median Coverage* | *% covered > 10x* | *% covered > 20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|
| ADCY3 | 126.4 | 99.9% | 98.8% | No OMIM disease ID |
| ANOS1 | 78.3 | 91.4% | 87.3% | Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 |
| BRCA2 | 103.2 | 99.7% | 98.8% | Wilms tumor, 194070<br>Fanconi anemia, complementation group D1, 605724 |
| CCDC141 | 110.4 | 99.8% | 97.8% | No OMIM Disease ID |
| CHD7 | 143.6 | 100.0% | 99.5% | CHARGE syndrome, 214800<br>Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 |
| DCC | 119.4 | 100.0% | 99.9% | Esophageal carcinoma, somatic, 133239<br>Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542<br>Mirror movements 1 and/or agenesis of the corpus callosum, 157600<br>Colorectal cancer, somatic, 114500 |
| DUSP6 | 184.5 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 |
| FEZF1 | 197.9 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 |
| FGF17 | 181.7 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 |
| FGF8 | 141.1 | 98.8% | 90.5% | Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 |
| FGFR1 | 131.6 | 100.0% | 99.7% | Pfeiffer syndrome, 101600<br>Jackson-Weiss syndrome, 123150<br>Trigonocephaly 1, 190440<br>Hypogonadotropic hypogonadism 2 with or without anosmia, 147950<br>Hartsfield syndrome, 615465<br>Osteoglophonic dysplasia, 166250<br>Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 |
| FLRT3 | 173.0 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 21 with anosmia, 615271 |
| FSHB | 111.9 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 24 without anosmia, 229070 |
| GNRH1 | 82.8 | 98.1% | 82.0% | ?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 |
| GNRHR | 148.9 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 7 without anosmia, 146110 |
| GPR161 | 181.5 | 100.0% | 100.0% | No OMIM Disease ID |

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| HESX1 | 65.7 | 99.9% | 97.5% | Pituitary hormone deficiency, combined, 5, 182230<br>Septooptic dysplasia, 182230<br>Growth hormone deficiency with pituitary anomalies, 182230 |
| HS6ST1 | 81.7 | 99.3% | 95.3% | No OMIM Disease ID |
| IGSF10 | 190.9 | 100.0% | 100.0% | No OMIM Disease ID |
| IL17RD | 143.4 | 99.9% | 99.3% | Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 |
| KISS1 | 90.8 | 100.0% | 99.1% | ?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 |
| KISS1R | 180.8 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 8 with or without anosmia, 614837<br>?Precocious puberty, central, 1, 176400 |
| KLB | 225.6 | 100.0% | 100.0% | No OMIM Disease ID |
| LEP | 184.9 | 100.0% | 100.0% | Obesity, morbid, due to leptin deficiency, 614962 |
| LEPR | 104.4 | 94.3% | 92.0% | Obesity, morbid, due to leptin receptor deficiency, 614963 |
| LHB | 25.3 | 95.0% | 61.7% | Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 |
| NR0B1 | 155.1 | 100.0% | 99.3% | Adrenal hypoplasia, congenital, 300200<br>46XY sex reversal 2, dosage-sensitive, 300018 |
| NSMF | 123.0 | 99.6% | 98.1% | Hypogonadotropic hypogonadism 9 with or without anosmia, 614838 |
| PALB2 | 146.3 | 100.0% | 99.9% | Fanconi anemia, complementation group N, 610832 |
| PCSK1 | 144.7 | 100.0% | 99.1% | Obesity with impaired prohormone processing, 600955 |
| PROK2 | 117.9 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 |
| PROKR2 | 245.3 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 |
| PROP1 | 107.5 | 93.9% | 86.2% | Pituitary hormone deficiency, combined, 2, 262600 |
| PTCH1 | 117.3 | 100.0% | 99.2% | Basal cell carcinoma, somatic, 605462<br>Basal cell nevus syndrome, 109400<br>Holoprosencephaly 7, 610828 |
| PTCH2 | 131.5 | 99.9% | 98.8% | Basal cell carcinoma, somatic, 605462<br>Basal cell nevus syndrome, 109400<br>Medulloblastoma, somatic, 155255 |
| SEMA3A | 154.6 | 100.0% | 100.0% | No OMIM disease ID |
| SMARCB1 | 192.9 | 100.0% | 100.0% | Rhabdoid tumors, somatic, 609322<br>Coffin-Siris syndrome 3, 614608 |
| SOX10 | 101.7 | 100.0% | 99.8% | Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584<br>PCWH syndrome, 609136<br>Waardenburg syndrome, type 4C, 613266 |
| SPRY4 | 187.2 | 100.0% | 100.0% | Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 |
| SUFU | 141.7 | 100.0% | 100.0% | Basal cell nevus syndrome, 109400<br>Medulloblastoma, desmoplastic, 155255<br>Joubert syndrome 32, 617757 |

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| TAC3 | 63.9 | 99.9% | 95.4% | Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 |
| TACR3 | 153.6 | 100.0% | 99.6% | Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 |
| TCF12 | 137.8 | 100.0% | 99.9% | Craniosynostosis 3, 615314 |
| TP53 | 96.8 | 99.9% | 99.0% | Breast cancer, somatic, 114480<br>Li-Fraumeni syndrome, 151623<br>Pancreatic cancer, somatic, 260350<br>Bone marrow failure syndrome 5, 618165<br>Nasopharyngeal carcinoma, somatic, 607107<br>Hepatocellular carcinoma, somatic, 114550 |
| WDR11 | 116.0 | 97.3% | 96.1% | 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 : December 11th , 2019.*
*This list is accurate for panel version DG 2.17*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*