HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 2.18 (40 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ADCY3	100%	99,10%	100%	100%	No OMIM disease ID
ANOS1	89,80%	88,90%	99,90%	99,40%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
CCDC141	100%	99,30%	100%	100%	No OMIM disease ID
CHD7	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
DCC	100%	100%	100%	100%	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500
DUSP6	100%	100%	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FEZF1	100%	99,90%	100%	100%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100%	100%	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	98,20%	88,90%	100%	99,60%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100%	99,90%	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FLRT3	100%	100%	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FSHB	100%	100%	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
GNRH1	100%	93,70%	100%	100%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100%	100%	100%	100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	99,70%	97,30%	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230

HS6ST1	92,90%	84,50%	100%	100%	No OMIM disease ID
IGSF10	100%	100%	100%	100%	No OMIM disease ID
IL17RD	99,90%	99,10%	100%	100%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KISS1	100%	98,30%	100%	100%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KLB	100%	99,90%	100%	100%	No OMIM disease ID
LEP	99,90%	97,30%	100%	100%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,30%	92,60%	94,60%	94,60%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	90,40%	38,90%	100%	100%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHX3	96,60%	96,50%	100%	100%	Pituitary hormone deficiency, combined, 3, 221750
NR0B1	100%	99,50%	100%	100%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NSMF	96,10%	95,60%	100%	100%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	100%	99,50%	100%	100%	Obesity with impaired prohormone processing, 600955
TAC3	100%	99,60%	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100%	100%	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
PLXNA1	100%	99,60%	100%	100%	No OMIM disease ID
PROK2	99,90%	98,50%	100%	100%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	92,60%	82,60%	100%	100%	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	100%	99,90%	100%	100%	No OMIM disease ID
SOX10	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX2	100%	100%	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SPRY4	100%	100%	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TCF12	100%	99,90%	100%	100%	Craniosynostosis 3, 615314
WDR11	98,00%	96,50%	100%	100%	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors