HYPOGONADOTROPIC HYPOGONADISM GENE PANEL DG 3.2.0 (43 genes)

Releasedate: 16-09-2021

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	100	100	No OMIM disease ID
ANOS1	89,8	88,3	99,9	99,4	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AXL	100	98,9	100	100	No OMIM disease ID
CCDC141	99,8	99,5	100	99,9	No OMIM disease ID
CHD7	100	99,2	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
DCC	100	100	100	100	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Esophageal carcinoma, somatic, 133239 Colorectal cancer, somatic, 114500 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DUSP6	100	100	100	100	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
FEZF1	100	100	100	100	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100	100	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	97,1	87,2	100	99,9	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100	99,3	100	100	Pfeiffer syndrome, 101600 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Hartsfield syndrome, 615465 Trigonocephaly 1, 190440 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	99,5	89,5	100	100	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841

GNRHR	100	100	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
HESX1	99,3	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HS6ST1	93,6	86,7	100	100	No OMIM disease ID
IGSF10	100	99,9	100	100	No OMIM disease ID
IL17RD	99,9	99	100	100	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
KISS1	100	98,2	100	100	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100	99,6	100	100	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KLB	100	99,9	100	100	No OMIM disease ID
LEP	100	99,6	100	100	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,1	92,3	94,6	94,5	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	91,7	42,8	100	100	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHX3	96,6	96,2	100	100	Pituitary hormone deficiency, combined, 3, 221750
NR0B1	99,9	99,2	100	100	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NSMF	96,9	95,5	100	100	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	99,9	99,4	100	100	Obesity with impaired prohormone processing, 600955
PLXNA1	100	99,9	100	100	No OMIM disease ID
POLG	99,9	98,8	100	100	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PROK2	99,9	98,9	100	100	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	91	80,2	100	100	Pituitary hormone deficiency, combined, 2, 262600
SEMA3A	100	99,7	100	100	No OMIM disease ID
SEMA3E	99,1	98,9	100	100	?CHARGE syndrome, 214800

SOX10	99,9	97,2	100	100	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX2	100	99,8	100	100	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SPRY4	100	100	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
TAC3	99,9	93,6	100	100	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100	100	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TCF12	99,9	99,7	100	100	Craniosynostosis 3, 615314
WDR11	98,2	96,5	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.
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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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} sign ify risk factors