Oslo universitetssykehus
Avdeling for medisinsk genetikk

Hypogonadotrop hypogonadisme

Genpanel, versjon v01

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet , gjenkjennes (https://genome.ucsc.edu).

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
BMP15	1068	NM_005448.2		1-2	Ovarian dysgenesis 2 OMIM Premature ovarian failure 4 OMIM
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM Hypogonadotropic hypogonadism 5 with or without anosmia OMIM
CUL4B	2555	NM_003588.3		2-22	Mental retardation, X-linked, syndromic 15 (Cabezas type) OMIM
CYP17A1	2593	NM_000102.3		1-8	17,20-lyase deficiency, isolated OMIM 17-alpha-hydroxylase/17,20-lyase deficiency OMIM
CYP19A1	2594	NM_031226.2		3-11	Aromatase deficiency OMIM Aromatase excess syndrome OMIM

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DCAF17	25784	NM_025000.3		1-14	Woodhouse-Sakati syndrome OMIM
DMXL2	2938	NM_015263.3		1-43	?Polyendocrine-polyneuropathy syndrome OMIM
DUSP6	3072	NM_001946.3		1-3	Hypogonadotropic hypogonadism 19 with or without anosmia OMIM
ESR1	3467	NM_000125.3		1-8	Estrogen resistance OMIM
FEZF1	22788	NM_001160264.2		1-5	Hypogonadotropic hypogonadism 22, with or without anosmia OMIM
FGF17	3673	NM_003867.3		1-5	Hypogonadotropic hypogonadism 20 with or without anosmia OMIM
FGF8	3686	NM_033163.3		1-6	Hypogonadotropic hypogonadism 6 with or without anosmia OMIM
FGFR1	3688	NM_023110.2		2-18	Hartsfield syndrome OMIM Hypogonadotropic hypogonadism 2 with or without anosmia OMIM Jackson-Weiss syndrome OMIM Osteoglophonic dysplasia OMIM Pfeiffer syndrome OMIM Trigonocephaly 1 OMIM
FLRT3	3762	NM_198391.2		3	Hypogonadotropic hypogonadism 21 with anosmia OMIM
FOXL2	1092	NM_023067.3		1	Blepharophimosis, epicanthus inversus, and ptosis, type 1 OMIM Blepharophimosis, epicanthus inversus, and ptosis, type 2 OMIM Premature ovarian failure 3 OMIM
FSHB	3964	NM_000510.2		2-3	Hypogonadotropic hypogonadism 24 without anosmia OMIM
FSHR	3969	NM_000145.3		1-10	Ovarian dysgenesis 1 OMIM Ovarian response to FSH stimulation OMIM
GLI2	4318	NM_005270.4		1-13	Culler-Jones syndrome OMIM Holoprosencephaly 9 OMIM

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GNRH1	4419	NM_000825.3		1-3	?Hypogonadotropic hypogonadism 12 with or without anosmia OMIM
GNRHR	4421	NM_000406.2		1-3	Hypogonadotropic hypogonadism 7 without anosmia OMIM
HAMP	15598	NM_021175.3		1-3	Hemochromatosis, type 2B OMIM
HESX1	4877	NM_003865.2		1-4	Growth hormone deficiency with pituitary anomalies OMIM Pituitary hormone deficiency, combined, 5 OMIM Septooptic dysplasia OMIM
HFE	4886	NM_000410.3		1-6	Hemochromatosis OMIM
HFE2	4887	NM_213653.3		2-4	Hemochromatosis, type 2A OMIM
HS6ST1	5201	NM_004807.2	2	1-2	{Hypogonadotropic hypogonadism 15 with or without anosmia} OMIM
IL17RD	17616	NM_017563.4		1-13	Hypogonadotropic hypogonadism 18 with or without anosmia OMIM
KAL1	6211	NM_000216.3	10-14	1-14	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1) OMIM
KISS1	6341	NM_002256.3		2-3	?Hypogonadotropic hypogonadism 13 with or without anosmia OMIM
KISS1R	4510	NM_032551.4		1-5	Hypogonadotropic hypogonadism 8 with or without anosmia OMIM
LEP	6553	NM_000230.2		2-3	Obesity, morbid, due to leptin deficiency OMIM
LEPR	6554	NM_002303.5		3-20	Obesity, morbid, due to leptin receptor deficiency OMIM
LHX4	21734	NM_033343.3		1-6	Pituitary hormone deficiency, combined, 4 OMIM
NR0B1	7960	NM_000475.4		1-2	46XY sex reversal 2, dosage-sensitive OMIM Adrenal hypoplasia, congenital OMIM

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NSMF	29843	NM_015537.4		1-15	Hypogonadotropic hypogonadism 9 with or without anosmia OMIM
PCSK1	8743	NM_000439.4		1-14	Obesity with impaired prohormone processing OMIM
PNPLA6	16268	NM_006702.4		3-35	Boucher-Neuhauser syndrome OMIM Oliver-McFarlane syndrome OMIM Spastic paraplegia 39, autosomal recessive OMIM
POLR3A	30074	NM_007055.3		1-31	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM
POR	9208	NM_000941.2		2-16	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis OMIM Disordered steroidogenesis due to cytochrome P450 oxidoreductase OMIM
PROK2	18455	NM_001126128.1		1-4	Hypogonadotropic hypogonadism 4 with or without anosmia OMIM
PROKR2	15836	NM_144773.3		2-3	Hypogonadotropic hypogonadism 3 with or without anosmia OMIM
PROP1	9455	NM_006261.4		1-3	Pituitary hormone deficiency, combined, 2 OMIM
RNF216	21698	NM_207111.3	2, 6-8	2-17	Cerebellar ataxia and hypogonadotropic hypogonadism OMIM
SEMA3A	10723	NM_006080.2		1-17	{Hypogonadotropic hypogonadism 16 with or without anosmia} OMIM
SLC29A3	23096	NM_018344.5		1-6	Histiocytosis-lymphadenopathy plus syndrome OMIM
SLC40A1	10909	NM_014585.5		1-8	Hemochromatosis, type 4 OMIM

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SOX10	11190	NM_006941.3		2-4	PCWH syndrome OMIM Waardenburg syndrome, type 2E, with or without neurologic involvement OMIM Waardenburg syndrome, type 4C OMIM
SOX2	11195	NM_003106.3	1	1	Microphthalmia, syndromic 3 OMIM Optic nerve hypoplasia and abnormalities of the central nervous system OMIM
SPRY4	15533	NM_030964.3		2-3	Hypogonadotropic hypogonadism 17 with or without anosmia OMIM
STUB1	11427	NM_005861.3		1-7	Spinocerebellar ataxia, autosomal recessive 16 OMIM
TAC3	11521	NM_013251.3		2-6	Hypogonadotropic hypogonadism 10 with or without anosmia OMIM
TACR3	11528	NM_001059.2		1-5	Hypogonadotropic hypogonadism 11 with or without anosmia OMIM
TFR2	11762	NM_003227.3		1-18	Hemochromatosis, type 3 OMIM
WDR11	13831	NM_018117.11		1-29	Hypogonadotropic hypogonadism 14 with or without anosmia OMIM