

Full name:	Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
Laboratory:	<u>Centre de Génétique Humaine - CHU Sart-Tilman</u>
Created:	12 Jul 2019 - 11:12
Changed:	25 Feb 2022 - 09:55

Related Diseases

- 46,XX ovotesticular difference of sex development
- 46,XX testicular difference of sex development
- 46,XY complete gonadal dysgenesis
- 46,XY partial gonadal dysgenesis
- Acrocallosal syndrome
- Adams-Oliver syndrome
- Alagille syndrome due to a JAG1 point mutation
- Alobar holoprosencephaly
- Autosomal recessive non-syndromic intellectual disability
- Autosomal thrombocytopenia with normal platelets
- Brachydactyly-arterial hypertension syndrome
- CHARGE syndrome
- Campomelic dysplasia
- Combined pituitary hormone deficiencies, genetic forms
- Endosteal sclerosis-cerebellar hypoplasia syndrome
- Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome
- Hypothyroidism due to deficient transcription factors involved in pituitary development or function
- Idiopathic achalasia
- Idiopathic central precocious puberty
- Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome
- Isolated follicle stimulating hormone deficiency
- Kallmann syndrome
- Leber congenital amaurosis
- Leydig cell hypoplasia due to LHB deficiency

- [Lobar holoprosencephaly](#)
- [Microform holoprosencephaly](#)
- [Midline interhemispheric variant of holoprosencephaly](#)
- [Non-acquired panhypopituitarism](#)
- [Non-seminomatous germ cell tumor of testis](#)
- [Normosmic congenital hypogonadotropic hypogonadism](#)
- [Obesity due to congenital leptin deficiency](#)
- [Obesity due to prohormone convertase I deficiency](#)
- [Omenn syndrome](#)
- [Pituitary stalk interruption syndrome](#)
- [Semilobar holoprosencephaly](#)
- [Septo-optic dysplasia spectrum](#)
- [Septopreoptic holoprosencephaly](#)
- [Tetralogy of Fallot](#)
- [WHIM syndrome](#)
- [X-linked adrenal hypoplasia congenita](#)
- [X-linked congenital generalized hypertrichosis](#)
- [X-linked intellectual disability with isolated growth hormone deficiency](#)

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
AMN1	99.52	0	NM_001113402
ANOS1	93.11	0	NM_000216
AXL	99.69	0	NM_021913
CCDC141	97.45	0	NM_173648
CCKBR	99.61	0	NM_176875
CHD7	99.53	0	NM_017780
CRY1	99.26	0	NM_004075
CXCR4	99.63	0	NM_003467

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DCC</u>	99.55	0	NM_005215
<u>DUSP6</u>	99.68	0	NM_001946
<u>FEZF1</u>	99.43	0	NM_001024613
<u>FGF13</u>	99.32	0	NM_004114
<u>FGF17</u>	99.67	0	NM_003867
<u>FGF8</u>	78.88	0	NM_033163
<u>FGFR1</u>	99.69	0	NM_023110
<u>FLRT3</u>	99.67	0	NM_198391
<u>FSHB</u>	99.64	0	NM_000510
<u>GAP43</u>	97.81	0	NM_001130064
<u>GLI3</u>	99.55	0	NM_000168
<u>GNRH1</u>	99.54	0	NM_000825
<u>GNRHR</u>	99.45	0	NM_000406
<u>HESX1</u>	98.36	0	NM_003865
<u>HS6ST1</u>	92.39	0	NM_004807
<u>IL17RD</u>	94.68	0	NM_017563
<u>JAG1</u>	97.49	0	NM_000214
<u>KISS1</u>	99.66	0	NM_002256
<u>KISS1R</u>	85.01	0	NM_032551
<u>KLB</u>	99.62	0	NM_175737

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>LEP</u>	99.69	0	NM_000230
<u>LEPR</u>	98.65	0	NM_002303
<u>LHB</u>	99.69	0	NM_000894
<u>MASTL</u>	99.20	0	NM_032844
<u>NOS1</u>	99.42	0	NM_000620
<u>NOTCH1</u>	96.74	0	NM_017617
<u>NR0B1</u>	99.34	0	NM_000475
<u>NRP2</u>	99.58	0	NM_201266
<u>NSMF</u>	93.42	0	NM_015537
<u>OTUD4</u>	98.02	0	NM_001102653
<u>PALM2</u>	98.64	0	NM_001037293
<u>PCSK1</u>	99.63	0	NM_000439
<u>PDE3A</u>	99.05	0	NM_000921
<u>PLEKHA5</u>	98.89	0	NM_001143821
<u>PLXNA1</u>	99.67	0	NM_032242
<u>POLR3B</u>	99.29	0	NM_018082
<u>PROK2</u>	90.75	0	NM_001126128
<u>PROKR2</u>	99.69	0	NM_144773
<u>PROP1</u>	99.69	0	NM_006261
<u>RD3</u>	99.63	0	NM_183059

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SEMA3A</u>	99.53	0	NM_006080
<u>SEMA3E</u>	99.45	0	NM_012431
<u>SEMA7A</u>	92.63	0	NM_003612
<u>SOX10</u>	93.26	0	NM_006941
<u>SOX2</u>	95.94	0	NM_003106
<u>SOX3</u>	83.29	0	NM_005634
<u>SOX9</u>	99.60	0	NM_001035235
<u>SPRY4</u>	99.69	0	NM_030964
<u>TAC3</u>	99.64	0	NM_013251
<u>TACR3</u>	99.41	0	NM_001059
<u>TRAPPC9</u>	99.57	0	NM_031466
<u>TSPAN11</u>	99.69	0	NM_001080509
<u>WDR11</u>	99.33	0	NM_018117