

Full name:	Thyroid dysgenesis (38 genes) - VUB
Description:	(in-silico subpanel of Mendeliome) https://www.brightcore.be/gene-panels/mendeliome---thyroid-disorders---v11 Kit used : Roche SeqCap EZ custom design
Type of panel:	<u>Custom panel</u>
Provider:	home made
Version number:	V11
Laboratory:	<u>Centrum Medische Genetica - UZ Brussel VUB</u>
Created:	17 Jul 2019 - 11:05
Changed:	01 Sep 2022 - 15:17

Related Diseases

- Athyreosis
- Autosomal dominant hypocalcemia
- Bartter syndrome with hypocalcemia
- Brain-lung-thyroid syndrome
- Cushing syndrome due to bilateral macronodular adrenocortical disease
- Familial gestational hyperthyroidism
- Familial hyperthyroidism due to mutations in TSH receptor
- Familial hypocalciuric hypercalcemia type 1
- Familial hypocalciuric hypercalcemia type 2
- Familial hypocalciuric hypercalcemia type 3
- Familial thyroid dysmorphogenesis
- Generalized resistance to thyroid hormone
- Genetic transient congenital hypothyroidism
- Hypothyroidism due to TSH receptor mutations
- Neonatal severe primary hyperparathyroidism
- Peripheral resistance to thyroid hormones

- Pituitary resistance to thyroid hormone
- Pseudohypoparathyroidism type 1A
- Pseudohypoparathyroidism type 1B
- Pseudohypoparathyroidism type 1C
- Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha
- Thyroid ectopia
- Thyroid hypoplasia

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>AP2S1</u>	100.00	0	No comment
<u>CASR</u>	100.00	0	No comment
<u>CDCA8</u>	100.00	0	No comment
<u>DUOX1</u>	100.00	0	No comment
<u>DUOX2</u>	100.00	0	No comment
<u>DUOXA2</u>	100.00	0	No comment
<u>DYRK1A</u>	100.00	0	No comment
<u>ELN</u>	100.00	0	No comment
<u>FOXE1</u>	60.48	0	No comment
<u>GAS1</u>	62.99	0	No comment
<u>GLIS3</u>	100.00	0	No comment
<u>GNA11</u>	99.51	0	No comment
<u>GNAS</u>	98.96	0	No comment
<u>GNAS-AS1</u>	100.00	0	No comment
<u>IYD</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>JAG1</u>	99.66	0	No comment
<u>KAT6B</u>	100.00	0	No comment
<u>KDM6A</u>	100.00	0	No comment
<u>KMT2D</u>	100.00	0	No comment
<u>NKX2-1</u>	92.46	0	No comment
<u>NKX2-5</u>	100.00	0	No comment
<u>NTN1</u>	95.84	0	No comment
<u>PAX8</u>	100.00	0	No comment
<u>SALL1</u>	100.00	0	No comment
<u>SECISBP2</u>	98.68	0	No comment
<u>SLC11A2</u>	100.00	0	No comment
<u>SLC26A4</u>	99.99	0	No comment
<u>SLC26A7</u>	100.00	0	No comment
<u>SLC5A5</u>	100.00	0	No comment
<u>STX16</u>	100.00	0	No comment
<u>TBX1</u>	77.39	0	No comment
<u>TG</u>	100.00	0	No comment
<u>THPO</u>	99.84	0	No comment
<u>THRA</u>	100.00	0	No comment
<u>THRB</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TSHR</u>	100.00	0	No comment
<u>TUBB1</u>	100.00	0	No comment
<u>URB1</u>	99.99	0	No comment