

Full name:	Early-onset severe obesity (44 genes) - ULG
Abbreviation:	OBZT panel
Description:	Early-onset severe obesity (monogenic or syndromic). Investigation of : - Point mutations in 44 genes panel by NGS - Deletions/duplications in 8 genes (LEP, LEPR, MC2R, MC3R, MC4R, POMC, SIM1, 16p11.2 region) by MLPA (MRC Holland P220)
Type of panel:	<u>Custom panel</u>
Provider:	Twist Biosciences
Laboratory:	<u>Centre de Génétique Humaine - CHU Sart-Tilman</u>
Created:	13 Dec 2019 - 14:56
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Related Diseases

- Abdominal obesity-metabolic syndrome 3
- Alström syndrome
- Autosomal dominant non-syndromic intellectual disability
- Bardet-Biedl syndrome
- Borjeson-Forssman-Lehmann syndrome
- Carpenter syndrome
- Congenital central hypoventilation syndrome
- Cushing syndrome due to bilateral macronodular adrenocortical disease
- Infantile spasms syndrome
- Joubert syndrome
- Joubert syndrome with hepatic defect
- Joubert syndrome with ocular defect
- Joubert syndrome with oculorenal defect
- MAGEL2-related Prader-Willi-like syndrome

- Meckel syndrome
- Non-specific syndromic intellectual disability
- Obesity due to SIM1 deficiency
- Obesity due to congenital leptin deficiency
- Obesity due to leptin receptor gene deficiency
- Obesity due to melanocortin 4 receptor deficiency
- Obesity due to pro-opiomelanocortin deficiency
- Obesity due to prohormone convertase I deficiency
- Rubinstein-Taybi syndrome due to 16p13.3 microdeletion
- Rubinstein-Taybi syndrome due to CREBBP mutations
- Rubinstein-Taybi syndrome due to EP300 haploinsufficiency
- Senior-Loken syndrome
- Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency
- Sotos syndrome
- Ulnar-mammary syndrome
- WAGR syndrome

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ADCY3</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>ALMS1</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>ARL6</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBIP1</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBS1</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBS10</u>	100.00	0	only for coding exons and intronic borders +/-14pb

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>BBS12</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBS2</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBS4</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBS5</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBS7</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>BBS9</u>	99.99	0	only for coding exons and intronic borders +/-14pb
<u>BDNF</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>CEP290</u>	99.99	0	only for coding exons and intronic borders +/-14pb
<u>CREBBP</u>	99.99	0	only for coding exons and intronic borders +/-14pb
<u>DYRK1B</u>	99.94	0	only for coding exons and intronic borders +/-14pb
<u>EP300</u>	99.99	0	only for coding exons and intronic borders +/-14pb
<u>GNAS</u>	99.34	0	only for coding exons and intronic borders +/-14pb

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>IFT27</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>INPP5E</u>	97.02	0	only for coding exons and intronic borders +/-14pb
<u>LEP</u>	100.00	1	only for coding exons and intronic borders +/-14pb
<u>LEPR</u>	99.99	1	only for coding exons and intronic borders +/-14pb
<u>LZTFL1</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>MAGEL2</u>	99.54	0	only for coding exons and intronic borders +/-14pb
<u>MC3R</u>	100.00	1	only for coding exons and intronic borders +/-14pb
<u>MC4R</u>	100.00	1	only for coding exons and intronic borders +/-14pb
<u>MKKS</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>MKS1</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>MRAP2</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>MYT1L</u>	100.00	0	only for coding exons and intronic borders +/-14pb

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>NTRK2</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>PCSK1</u>	99.99	0	only for coding exons and intronic borders +/-14pb
<u>PHF6</u>	99.98	0	only for coding exons and intronic borders +/-14pb
<u>POMC</u>	99.97	1	only for coding exons and intronic borders +/-14pb
<u>RAB23</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>SDCCAG8</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>SETD2</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>SH2B1</u>	100.00	1	only for coding exons and intronic borders +/-14pb
<u>SIM1</u>	100.00	1	only for coding exons and intronic borders +/-14pb
<u>TBX3</u>	98.58	0	only for coding exons and intronic borders +/-14pb
<u>TRIM32</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>TTC8</u>	100.00	0	only for coding exons and intronic borders +/-14pb

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TUB</u>	100.00	0	only for coding exons and intronic borders +/-14pb
<u>WDPCP</u>	100.00	0	only for coding exons and intronic borders +/-14pb