

Full name:	Brain malformations (34 genes) - ULB
Laboratory:	<u>Centre de Génétique Humaine - Erasme ULB</u>
Created:	09 Dec 2019 - 12:32
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Related Diseases

- Autosomal recessive primary microcephaly
- Ear-patella-short stature syndrome
- Microcephalic osteodysplastic primordial dwarfism type II
- Microform holoprosencephaly
- Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome
- Seckel syndrome

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ASPM</u>	98.88	0	
<u>CCDC88C</u>	100.00	0	
<u>CDK5RAP2</u>	99.64	0	
<u>CDON</u>	100.00	0	
<u>CENPJ</u>	100.00	0	
<u>CEP135</u>	100.00	0	
<u>CEP152</u>	99.96	0	
<u>CRIPTO</u>	100.00	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DISP1</u>	100.00	0	
<u>DLL1</u>	100.00	0	
<u>FGF8</u>	99.79	0	
<u>FGFR1</u>	94.19	0	
<u>FOXH1</u>	100.00	0	
<u>GAS1</u>	100.00	0	
<u>GCM2</u>	78.95	0	
<u>GLI2</u>	100.00	0	
<u>L1CAM</u>	96.28	0	
<u>MCPH1</u>	99.98	0	
<u>MPDZ</u>	99.70	0	
<u>NODAL</u>	100.00	0	
<u>ORC4</u>	100.00	0	
<u>ORC6</u>	100.00	0	
<u>PCNT</u>	100.00	0	
<u>PTCH1</u>	99.88	0	
<u>SHH</u>	98.82	0	
<u>SIX3</u>	99.47	0	
<u>SLC25A15</u>	99.87	0	
<u>SMAD2</u>	99.61	0	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>STIL</u>	100.00	0	
<u>SUFU</u>	100.00	0	
<u>TGIF1</u>	100.00	0	
<u>TRMT10A</u>	100.00	0	
<u>WDR62</u>	100.00	0	
<u>ZIC2</u>	100.00	0	