

Full name:	Overgrowth (24 genes) - IPG
Version number:	version 3
Laboratory:	<u>Centre de Génétique-Institut de Pathologie et de Génétique (IPG)</u>
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Related Diseases

- 5q35 microduplication syndrome
- Beckwith-Wiedemann syndrome due to CDKN1C mutation
- Beckwith-Wiedemann syndrome due to NSD1 mutation
- CLOVES syndrome
- Cowden syndrome
- Deletion 5q35
- Hemimegalencephaly
- IMAGe syndrome
- Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome
- Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome
- Malan overgrowth syndrome
- Marshall-Smith syndrome
- Megalencephaly-capillary malformation-polymicrogyria syndrome
- Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome
- Proteus syndrome
- Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome
- Segmental progressive overgrowth syndrome with fibroadipose hyperplasia
- Silver-Russell syndrome due to a point mutation
- Simpson-Golabi-Behmel syndrome
- Sotos syndrome
- Tatton-Brown-Rahman syndrome
- Weaver syndrome

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ADARB2</u>	100.00	1	NM_018702.4
<u>AKT1</u>	100.00	1	NM_001014431.2
<u>AKT3</u>	100.00	1	NM_005465.7
<u>BRWD3</u>	100.00	1	NM_153252.5
<u>CDKN1C</u>	100.00	1	NM_001362474.2
<u>CHD3</u>	100.00	1	NM_001005273.3
<u>CHD8</u>	100.00	1	NM_001170629.2
<u>DIS3L2</u>	100.00	1	NM_152383.4
<u>DNMT3A</u>	100.00	1	NM_175629.2
<u>EED</u>	100.00	1	NM_003797.5
<u>EZH2</u>	100.00	1	NM_004456.5
<u>GPC3</u>	100.00	1	NM_001164617.2
<u>H1-4</u>	100.00	1	NM_005321.3
<u>KMT5B</u>	100.00	1	NM_017635.5
<u>MTOR</u>	100.00	1	NM_004958.4
<u>NFIX</u>	100.00	1	NM_002501.4
<u>NSD1</u>	100.00	1	NM_022455.4
<u>PDGFRB</u>	100.00	1	NM_002609.4
<u>PIK3CA</u>	100.00	1	NM_006218.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PPP2R5C</u>	100.00	1	NM_001161725.1
<u>PPP2R5D</u>	100.00	1	NM_006245.4
<u>PTEN</u>	100.00	1	NM_000314.8
<u>SETD2</u>	100.00	1	NM_014159.6
<u>TCF20</u>	100.00	1	NM_005650.3