

Full name:	Autism (57 genes) - IPG
Version number:	Version 3
Laboratory:	<u>Centre de Génétique-Institut de Pathologie et de Génétique (IPG)</u>
Created:	04 Jul 2019 - 08:45
Changed:	10 Oct 2022 - 14:50

Related Diseases

- Fragile X 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>ADNP</u>	100.00	1	NM_001282531.3
<u>ANK2</u>	100.00	1	NM_001148.6
<u>ANKRD11</u>	100.00	1	NM_013275.6
<u>ARID1B</u>	98.00	1	NM_001374828.1
<u>BCKDK</u>	100.00	1	NM_005881.4
<u>CACNA1C</u>	100.00	1	NM_000719.7
<u>CHD3</u>	100.00	1	NM_001005273.3
<u>CHD7</u>	100.00	1	NM_017780.4
<u>CHD8</u>	100.00	1	NM_001170629.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CHRNA7</u>	0.00	1	NM_000746.6 / Exons 1-4 (5-10 not covered) / % unknown
<u>CNTNAP2</u>	100.00	1	NM_014141.6
<u>CNTNAP4</u>	100.00	1	NM_033401.5
<u>DHCR7</u>	100.00	1	NM_001360.3
<u>DYRK1A</u>	100.00	1	NM_001347721.2
<u>FMR1</u>	100.00	1	NM_002024.6
<u>FOXP1</u>	100.00	1	NM_032682.6
<u>FOXP2</u>	100.00	1	NM_014491.4
<u>GRIN2A</u>	100.00	1	NM_001134407.3
<u>GRIN2B</u>	100.00	1	XM_011520629.2
<u>HDAC4</u>	100.00	1	NM_006037.3
<u>KATNAL2</u>	100.00	1	NM_031303.3
<u>KCNB1</u>	100.00	1	NM_004975.4
<u>MECP2</u>	100.00	1	NM_001110792.2 and NM_004992.3
<u>MEF2C</u>	100.00	1	NM_002397.5
<u>MTOR</u>	100.00	1	NM_004958.4
<u>NIPBL</u>	100.00	1	NM_133433.4
<u>NLGN1</u>	100.00	1	NM_014932.5
<u>NLGN3</u>	100.00	1	NM_181303.2

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<u>NLGN4X</u>	100.00	1	NM_181332.3
<u>NLGN4Y</u>	100.00	1	NM_014893.4
<u>NRXN1</u>	100.00	1	NM_001330078.2
<u>NRXN2</u>	100.00	1	NM_015080.4
<u>NRXN3</u>	100.00	1	NM_001330195.2
<u>POGZ</u>	100.00	1	NM_015100.4
<u>PPP2R5D</u>	100.00	1	NM_006245.4
<u>PTCHD1</u>	100.00	1	NM_173495.3
<u>PTEN</u>	100.00	1	NM_000314.8
<u>RAI1</u>	100.00	1	NM_030665.4
<u>RIMS1</u>	100.00	1	NM_014989.5
<u>SCN1A</u>	100.00	1	NM_001165963.3
<u>SCN2A</u>	100.00	1	NM_001040142.2
<u>SCN8A</u>	100.00	1	NM_001330260.2
<u>SHANK1</u>	100.00	1	NM_016148.5
<u>SHANK2</u>	100.00	1	NM_133266.5
<u>SHANK3</u>	100.00	1	NM_001372044.1
<u>SMC1A</u>	100.00	1	NM_006306.4
<u>SMC3</u>	100.00	1	NM_005445.4
<u>SYNGAP1</u>	100.00	1	NM_006772.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TBL1XR1</u>	100.00	1	NM_024665.7
<u>TBR1</u>	100.00	1	NM_006593.4
<u>TCF20</u>	100.00	1	NM_005650.3
<u>TMLHE</u>	100.00	1	NM_018196.4
<u>TSC1</u>	100.00	1	NM_000368.5
<u>TSC2</u>	100.00	1	NM_000548.5
<u>UBE3A</u>	100.00	1	NM_130839.5
<u>VPS13B</u>	100.00	1	NM_017890.4