

Full name:	Intellectual Disability (104 genes) - IPG
Version number:	Version 3
Laboratory:	<u>Centre de Génétique-Institut de Pathologie et de Génétique (IPG)</u>
Created:	20 Dec 2021 - 12:50
Changed:	10 Oct 2022 - 15:13

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ADNP</u>	100.00	1	NM_001282531.3
<u>ANKRD11</u>	100.00	1	NM_013275.6
<u>ARHGEF9</u>	100.00	1	NM_001353921.2
<u>ARID1A</u>	100.00	1	NM_006015.6
<u>ARID1B</u>	98.83	1	NM_001374828.1
<u>ARX</u>	100.00	1	NM_139058.3
<u>ASXL3</u>	100.00	1	NM_030632.3
<u>ATRX</u>	100.00	1	NM_000489.5
<u>BCL11A</u>	100.00	1	NM_022893.4
<u>BRAF</u>	100.00	1	NM_001354609.2
<u>BRWD3</u>	100.00	1	NM_153252.5
<u>CAMK2G</u>	100.00	1	NM_172171.2
<u>CAMTA1</u>	100.00	1	NM_015215.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CASK</u>	100.00	1	NM_003688.3
<u>CBL</u>	100.00	1	NM_005188.4
<u>CDKL5</u>	100.00	1	NM_001323289.2
<u>CERT1</u>	100.00	1	NM_001130105.1
<u>CHAMP1</u>	100.00	1	NM_032436.4
<u>CHD2</u>	100.00	1	NM_001271.4
<u>CNOT3</u>	100.00	1	NM_014516.4
<u>CREBBP</u>	100.00	1	NM_004380.3
<u>CTNNB1</u>	100.00	1	NM_001904.4
<u>CTSA</u>	100.00	1	NM_000308.4
<u>DDX3X</u>	100.00	1	NM_001356.4
<u>DHCR7</u>	100.00	1	NM_001360.3
<u>DNM1</u>	100.00	1	NM_004408.4
<u>DYRK1A</u>	100.00	1	NM_001347721.2
<u>EHMT1</u>	100.00	1	NM_024757.5
<u>EP300</u>	100.00	1	NM_001429.4
<u>EZH2</u>	100.00	1	NM_004456.5
<u>FGD1</u>	100.00	1	NM_004463.3
<u>FGFR3</u>	100.00	1	NM_001163213.1
<u>FMR1</u>	100.00	1	NM_002024.6

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>FOXG1</u>	100.00	1	NM_005249.5
<u>FOXP1</u>	100.00	1	NM_032682.6
<u>GLI3</u>	100.00	1	NM_000168.6
<u>GNAO1</u>	100.00	1	NM_020988.3
<u>HACE1</u>	100.00	1	NM_020771.4
<u>HIVEP2</u>	100.00	1	NM_006734.4
<u>HNRNPK</u>	100.00	1	NM_031263.4
<u>HNRNPU</u>	100.00	1	NM_031844.3
<u>HPRT1</u>	100.00	1	NM_000194.3
<u>HRAS</u>	100.00	1	NM_005343.4
<u>HUWE1</u>	100.00	1	NM_031407.7
<u>IQSEC2</u>	100.00	1	NM_001111125.3
<u>ITPR1</u>	100.00	1	NM_001168272.1
<u>KANSL1</u>	100.00	1	NM_001193466.2
<u>KAT6A</u>	100.00	1	NM_006766.5
<u>KCNH1</u>	100.00	1	NM_172362.3
<u>KCNQ2</u>	100.00	1	NM_172107.4
<u>KDM6A</u>	100.00	1	NM_001291415.2
<u>KIF1A</u>	100.00	1	NM_001244008.1
<u>KMT2A</u>	100.00	1	NM_001197104.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>KPTN</u>	100.00	1	NM_007059.4
<u>KRAS</u>	100.00	1	NM_033360.4
<u>LRP1</u>	100.00	1	NM_002332.3
<u>MECP2</u>	100.00	1	NM_001110792.2 et NM_004992.3
<u>MED12</u>	100.00	1	NM_005120.3
<u>MED13L</u>	100.00	1	NM_015335.4
<u>MID1</u>	100.00	1	NM_000381.4
<u>MTOR</u>	100.00	1	NM_004958.4
<u>NAA10</u>	100.00	1	NM_003491.4
<u>NALCN</u>	100.00	1	NM_052867.4
<u>NEXMIF</u>	100.00	1	NM_001008537.3
<u>NF1</u>	100.00	1	NM_001042492.3
<u>NFIX</u>	100.00	1	NM_002501.4
<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
<u>NLGN4X</u>	100.00	1	NM_181332.3
<u>NSD1</u>	100.00	1	NM_022455.4
<u>PACS1</u>	100.00	1	NM_018026.4
<u>PCGF2</u>	100.00	1	NM_007144.3

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<u>PIGT</u>	100.00	1	NM_015937.6
<u>POU1F1</u>	100.00	1	NM_000306.4
<u>PPP2R1A</u>	100.00	1	NM_014225.6
<u>PPP2R5D</u>	100.00	1	NM_006245.4
<u>PTCHD1</u>	100.00	1	NM_173495.3
<u>PURA</u>	100.00	1	NM_005859.5
<u>RAI1</u>	100.00	1	NM_030665.4
<u>SATB2</u>	100.00	1	NM_001172509.2
<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>SETBP1</u>	100.00	1	NM_015559.3
<u>SETD5</u>	100.00	1	NM_001080517.3
<u>SHANK1</u>	100.00	1	NM_016148.5
<u>SHANK2</u>	100.00	1	NM_133266.5
<u>SLC2A1</u>	100.00	1	NM_006516.3
<u>SLC9A6</u>	100.00	1	NM_001042537.1
<u>SMARCA2</u>	100.00	1	NM_003070.5
<u>SMC1A</u>	100.00	1	NM_006306.4
<u>SMC3</u>	100.00	1	NM_005445.4

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<u>SMS</u>	100.00	1	NM_004595.5
<u>SOX3</u>	100.00	1	NM_005634.2
<u>STXBP1</u>	100.00	1	NM_001032221.6
<u>SYNGAP1</u>	100.00	1	NM_006772.3
<u>TBR1</u>	100.00	1	NM_006593.4
<u>TCF4</u>	100.00	1	NM_001083962.2
<u>TUBB3</u>	100.00	1	NM_006086.4
<u>UBE3A</u>	100.00	1	NM_130839.5
<u>USP9X</u>	100.00	1	NM_001039591.3
<u>VPS13B</u>	100.00	1	NM_017890.4
<u>WDR45</u>	100.00	1	NM_001029896.2