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Full name:	Dystonia (68 genes) - KUL
Version number:	V5
Laboratory:	Centrum Menselijke Erfelijkheid - KUL
Created:	16 Dec 2022 - 10:34
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Related Diseases

- Adult-onset cervical dystonia, DYT23 type
- Ataxia-telangiectasia
- Autosomal dominant dopa-responsive dystonia
- Autosomal recessive dopa-responsive dystonia
- Bethlem myopathy
- Classic pantothenate kinase-associated neurodegeneration
- Cranio-cervical dystonia with laryngeal and upper-limb involvement
- Developmental malformations-deafness-dystonia syndrome
- Dopa-responsive dystonia due to sepiapterin reductase deficiency
- Familial dyskinesia and facial myokymia
- Infantile spasms syndrome
- MEPAN syndrome
- Mohr-Tranebjaerg syndrome
- Niemann-Pick disease type C, adult neurologic onset
- Niemann-Pick disease type C, severe perinatal form
- Partington syndrome
- Primary dystonia, DYT27 type
- Primary dystonia, DYT4 type
- Primary dystonia, DYT6 type
- Sandhoff disease, adult form
- Sandhoff disease, infantile form
- Sandhoff disease, juvenile form
- Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome

- Tay-Sachs disease, B variant, adult form
- Wilson disease

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ACTB	100.00	0	NM_001101.4
ADCY5	98.60	0	NM_183357.2
ANO3	99.70	0	NM_031418.3
ARSA	100.00	0	NM_000487.5
ARX	96.10	0	NM_139058.2
ATM	100.00	0	NM_000051.3
ATP1A3	100.00	0	NM_152296.5
ATP7B	100.00	0	NM_000053.3
AUH	100.00	0	NM_001698.2
BCAP31	100.00	0	NM_001139441.1
C19ORF12	100.00	0	NM_001031726.3
CACNA1A	100.00	0	NM_002143.2
CACNA1B	97.90	0	NM_000718.3
CIZ1	98.80	0	NM_012127.2
COASY	100.00	0	NM_025233.6
COL6A3	100.00	0	NM_004369.3
CP	99.50	0	NM_000096.3
CYP27A1	100.00	0	NM_000784.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
DCAF17	100.00	0	NM_025000.3
DDC	100.00	0	NM_000790.3
DRD2	100.00	0	NM_000795.3
FA2H	99.90	0	NM_024306.4
FTL	100.00	0	NM_000146.3
GCDH	99.90	0	NM_000159.3
GCH1	100.00	0	NM_000161.2
GLB1	100.00	0	NM_000404.3
GNAL	100.00	0	NM_001142339.2
GNAO1	100.00	0	NM_020988.2
GNB1	100.00	0	NM_002074.4
HEXA	100.00	0	NM_000520.5
HEXB	100.00	0	NM_000521.3
HPRT1	100.00	0	NM_000194.2
KCNMA1	100.00	0	NM_002247.3
KCTD17	100.00	0	NM_001282684.1
KMT2B	98.40	0	NM_014727.2
MECR	100.00	0	NM_016011.4
MR1	100.00	0	NM_001194999.1
NKX2-1	100.00	0	NM_001079668.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
NPC1	99.90	0	NM_000271.4
NPC2	100.00	0	NM_006432.3
PANK2	100.00	0	NM_153638.3
PLA2G6	99.70	0	NM_003560.3
PNKD	100.00	0	NM_015488.4
PRKRA	93.20	0	NM_003690.4
PRRT2	99.60	0	NM_145239.2
PTS	100.00	0	NM_000317.2
RELN	100.00	0	NM_005045.3
SERAC1	100.00	0	NM_032861.3
SGCE	100.00	0	NM_003919.2
SLC18A2	100.00	0	NM_003054.4
SLC19A3	100.00	0	NM_025243.3
SLC2A1	100.00	0	NM_006516.2
SLC30A10	100.00	0	NM_018713.2
SLC39A14	100.00	0	NM_015359.5
SLC6A3	100.00	0	NM_001044.4
SMPD1	99.30	0	NM_000543.4
SPR	99.90	0	NM_003124.4
SUCLG1	99.60	0	NM_003849.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
TAF1	99.90	0	NM_004606.4
TBCD	99.70	0	NM_005993.4
TH	99.10	0	NM_199292.2
THAP1	100.00	0	NM_018105.2
TIMM8A	100.00	0	NM_004085.3
TOR1A	99.70	0	NM_000113.2
TUBB4A	100.00	0	NM_006087.3
VAC14	100.00	0	NM_018052.4
VPS13A	100.00	0	NM_033305.2
WDR45	100.00	0	NM_007075.3