DISEASE:
Presynaptic congenital myasthenic syndromes

NAME:	Presynaptic congenital myasthenic syndromes
ORPHACODE:	98914
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	CHAT AGRN SLC25A1 SNAP25 SYT2 MYO9A SLC5A7 VAMP1 SLC18A3 COL13A1
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RELATED CONTENT

Related Genetic Tests

- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- agrin
- choline O-acetyltransferase
- collagen type XIII alpha 1 chain
- myosin IXA
- solute carrier family 18 member A3
- solute carrier family 25 member 1
- solute carrier family 5 member 7
- synaptosome associated protein 25
- synaptotagmin 2
- vesicle associated membrane protein 1

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB
