ANALYTE:
TRPV4

NAME:	transient receptor potential cation channel subfamily V member 4
SYMBOL:	TRPV4
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	CMT2C OTRPC4 TRP12 VR-OAC VRL-2 VROAC osmosensitive transient receptor potential channel 4
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Hereditary spastic paraplegia (gene panel - 249 genes)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Myopathy (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Neuromuscular disorders (548 genes)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- Neuropathy (gene panel)
- Neuropathy (gene panel)
- Peripheral neuropathy (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)

Related Diseases

- Autosomal dominant Charcot-Marie-Tooth disease type 2C
- Autosomal dominant brachyolmia
- Autosomal dominant congenital benign spinal muscular atrophy

- Familial avascular necrosis of femoral head
- Familial digital arthropathy-brachydactyly
- Metatropic dysplasia
- Parastremmatic dwarfism
- Scapuloperoneal spinal muscular atrophy
- Spondyloepiphyseal dysplasia, Maroteaux type
- Spondylometaphyseal dysplasia, Kozlowski type

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Hereditary spastic paraplegia (188 genes) - ULB
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Myopathy (332 genes) - IPG
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Neuromuscular disorders (548 genes) - ULB
- Neuromuscular disorders (166 genes) - VUB
- Neuropathy (148 genes) - IPG
- Neuropathy (genepanel) - UZA
- Neuropathy panel - UGent
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent
