DISEASE:
Autosomal dominant congenital benign spinal muscular atrophy

NAME:	Autosomal dominant congenital benign spinal muscular atrophy
DESCRIPTION:	A rare distal hereditary motor neuropathy, with a variable clinical phenotype, typically characterized by congenital, non-progressive, predominantly distal, lower limb muscle weakness and atrophy and congenital (or early-onset) flexion contractures of the hip, knee and ankle joints. Reduced or absent lower limb deep tendon reflexes, skeletal anomalies (bilateral talipes equinovarus, scoliosis, kyphoscoliosis, lumbar hyperlordisis), late ambulation, waddling gait, joint hyperlaxity and/or bladder and bowel dysfuntion are usually also associated.
ORPHACODE:	1216
SYNONYMS:	Autosomal dominant benign distal spinal muscular atrophy Congenital benign spinal muscular atrophy with contractures Congenital nonprogressive spinal muscular atrophy
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	TRPV4
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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)
- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- transient receptor potential cation channel subfamily V member 4

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB
- Neuropathy (148 genes) - IPG