DISEASE:
Congenital myopathy, Paradas type

NAME:	Congenital myopathy, Paradas type
DESCRIPTION:	A rare congenital muscular dystrophy characterized by early onset of hypotonia, delayed motor development, and variably progressive generalized muscle weakness. Predominant involvement of pelvic and neck flexor muscles has been reported, as well as early involvement of hamstrings and medial gastrocnemius visible on muscle MRI. Serum creatine kinase levels are markedly elevated (in some cases already from early childhood). Muscle biopsy shows absence of dysferlin.
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