

DISEASE:
Steinert myotonic dystrophy

NAME:	Steinert myotonic dystrophy
DESCRIPTION:	A rare genetic multi-system disorder characterized by a wide range of muscle-related manifestations (muscle weakness, myotonia, early onset cataracts (before age 50) and systemic manifestations (cerebral, endocrine, cardiac, gastrointestinal tract, uterus, skin and immunologic involvement) that vary depending on the age of onset. The very wide clinical spectrum ranges from lethal presentations in infancy to mild, late-onset disease.
ORPHACODE:	273
SYNONYMS:	Myotonic dystrophy type 1 Steinert disease
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	DMPK
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