

DISEASE:**Epidermolysis bullosa simplex with muscular dystrophy**

NAME:	Epidermolysis bullosa simplex with muscular dystrophy
DESCRIPTION:	A form of epidermolysis bullosa simplex (EBS) characterized by generalized blistering associated with muscular dystrophy.
ORPHACODE:	257
SYNONYMS:	EBS with muscular dystrophy EBS-MD Limb-girdle muscular dystrophy with epidermolysis bullosa simplex
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>PLEC</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/1596>

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

- plectin

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB

Source URL: <http://gentest.healthdata.be/disease/1596>