

DISEASE:
AGel amyloidosis

NAME:	AGel amyloidosis
DESCRIPTION:	A rare, systemic amyloidosis characterized by a triad of ophthalmologic, neurologic and dermatologic findings due to the deposition of gelsolin amyloid fibrils in these tissues. Clinical manifestations include corneal lattice dystrophy, cranial neuropathy, especially affecting the facial nerve, bulbar signs, cutis laxa, increased skin fragility, and less commonly peripheral neuropathy and renal failure.
ORPHACODE:	85448
SYNONYMS:	Familial amyloid polyneuropathy type IV Familial amyloidosis, Finnish type Gelsolin amyloidosis Hereditary amyloidosis, Finnish type
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GSN
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/3342>

RELATED CONTENT

Related Genetic Tests

- [Corneal dystrophy \(gene panel\)](#)

Related Laboratories

- [Centrum Medische Genetica - UZ Gent](#)

Related Analytes

- [gelsolin](#)

Related Gene Panels

- [Corneal dystrophy - UGent](#)

Source URL: <http://gentest.healthdata.be/disease/3342>