DISEASE:
Autosomal dominant epidermolytic ichthyosis

NAME:	Autosomal dominant epidermolytic ichthyosis
DESCRIPTION:	A rare keratinopathic ichthyosis (KPI) characterized by a blistering phenotype at birth which progressively becomes hyperkeratotic.
ORPHACODE:	312
SYNONYMS:	BCIE Bullous congenital ichthyosiform erythroderma Bullous congenital ichthyosiform erythroderma of Brock Bullous ichthyosis EHK EI Epidermolytic hyperkeratosis Ichthyosis hystrix Brocq type
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	KRT1 KRT10

CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ichthyosis (gene panel)
- Keratinopathic ichthyosis (epidermolytic ichtyosis, superficial epidermolytic ichthyosis, congenital reticular ichthyosiform erythroderma) (3 genes)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- keratin 1
- keratin 10

Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL
- keratinopathic ichthyosis (3 genes) - KUL