DISEASE:
Harlequin ichthyosis

NAME:	Harlequin ichthyosis
DESCRIPTION:	A rare autosomal recessive congenital ichthyosis characterized at birth by the presence of large, thick, plate-like shell over the whole body associated with severe ectropion, eclabium, and flattened ears, that later develops into a severe scaling erythroderma. Harlequin ichthyosis is the most severe disorder of this group.
ORPHACODE:	457
SYNONYMS:	Autosomal congenital ichthyosis, Harlequin type HI Ichthyosis congenita, Harlequin type
XREF(S):	Orphanet OMIM ICD-10 MedDRA
ANALYTE(S):	ABCA12
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