DISEASE:
Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome

NAME:	Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome
DESCRIPTION:	Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome is a rare, genetic, corneal dystrophy disorder characterized by corneal opacification and dyskeratosis (which may cause visual impairment), associated with systemic features including palmoplantar hyperkeratosis, laryngeal dyskeratosis, pruritic hyperkeratotic scars, chronic rhintis, dyshidrosis and/or nail thickening.
ORPHACODE:	352662
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	NLRP1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Corneal dystrophy (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- NLR family pyrin domain containing 1

Related Gene Panels

- Corneal dystrophy - UGent