DISEASE:
Dyskeratosis congenita

NAME:	Dyskeratosis congenita
DESCRIPTION:	A rare ectodermal dysplasia syndrome that often presents with the classic triad of nail dysplasia, skin pigmentary changes, and oral leukoplakia associated with a high risk of bone marrow failure (BMF) and cancer.
ORPHACODE:	1775
SYNONYMS:	DC DKC Zinsser-Engman-Cole syndrome

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM MeSH MedDRA ICD-10
ANALYTE(S):	NPM1 DKC1 TERT TERC TINF2 NOP10 NHP2 USB1 WRAP53 CTC1 RTEL1 PARN TYMS
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Dyskeratosis Congenita (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- CST telomere replication complex component 1
- dyskerin pseudouridine synthase 1
- NHP2 ribonucleoprotein
- NOP10 ribonucleoprotein
- nucleophosmin 1
- poly(A)-specific ribonuclease
- regulator of telomere elongation helicase 1
- telomerase RNA component
- telomerase reverse transcriptase
- TERF1 interacting nuclear factor 2
- thymidylate synthetase
- U6 snRNA biogenesis phosphodiesterase 1
- WD repeat containing antisense to TP53

Related Gene Panels

- Dyskeratosis Congenita (18 genes) - KUL

Source URL: http://gentest.healthdata.be/disease/298