

DISEASE:
Palmoplantar keratoderma-deafness syndrome

NAME:	Palmoplantar keratoderma-deafness syndrome
DESCRIPTION:	Palmoplantar keratoderma-deafness syndrome is a keratinization disorder characterized by focal or diffuse palmoplantar keratoderma. A patchy distribution is observed with accentuation on the thenars, hypothenars and the arches of the feet. The disease becomes apparent in infancy and is associated with sensorineural hearing loss that shows a variable age of onset. Due to genetic and clinical similarities, it has been proposed that palmoplantar keratoderma-deafness syndrome, knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome and keratoderma hereditarium mutilans may represent variants of one broad disorder of syndromic deafness with heterogeneous phenotype. The disease is transmitted in an autosomal dominant manner with incomplete penetrance.
ORPHACODE:	2202
SYNONYMS:	PPK-deafness syndrome Palmoplantar hyperkeratosis-deafness syndrome Palmoplantar hyperkeratosis-hearing loss syndrome Palmoplantar keratoderma-hearing loss syndrome
XREF(S):	Orphanet MeSH OMIM ICD-10

ANALYTE(S):	<u>GJB2</u> <u>MT-TS1</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/918>

RELATED CONTENT

Related Genetic Tests

- [Epidermolysis bullosa \(gene panel\)](#)
- [Ichthyosis \(gene panel\)](#)

Related Laboratories

- [Centrum Menselijke Erfelijkheid - KUL](#)

Related Analytes

- [gap junction protein beta 2](#)
- [mitochondrially encoded tRNA-Ser \(UCN\) 1](#)

Related Gene Panels

- [Epidermolysis bullosa and bladder diseases \(60 genes\) - KUL](#)
- [Ichthyosis and erythroderma \(98 genes\) - KUL](#)

Source URL: <http://gentest.healthdata.be/disease/918>