DISEASE:
Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome

NAME:	Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome
DESCRIPTION:	A rare, syndromic genetic deafness disease characterized by symmetric or asymmetric knuckle pads (typically located on the distal and interphalangeal joints), leukonychia, diffuse palmoplantar keratoderma, and congenital, mild to moderate sensorineural deafness.
ORPHACODE:	2698
SYNONYMS:	Bart-Pumphrey syndrome Knuckle pads-leukonychia-sensorineural deafness-palmoplantar keratoderma syndrome Knuckle pads-leukonychia-sensorineural hearing loss-palmoplantar hyperkeratosis syndrome Knuckle pads-leukonychia-sensorineural hearing loss-palmoplantar keratoderma syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GJB2
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