DISEASE:
Deafness-lymphedema-leukemia syndrome

NAME:	Deafness-lymphedema-leukemia syndrome
DESCRIPTION:	A rare genetic disease characterized by the association of primary lymphedema (typically presenting in one or both lower limbs and frequently affecting the genitalia) and acute myeloid leukemia (often preceded by pancytopenia or myelodysplasia), with or without congenital deafness. Additional reported features include bilateral syndactyly of the toes, hypotelorism and epicanthic folds, long tapering fingers, and neck webbing.
ORPHACODE:	3226
SYNONYMS:	Emberger syndrome Hearing loss-lymphedema-leukemia syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GATA2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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