Opis choroby *

Definicja

A rare ectodermal dysplasia syndrome characterized by linear hypopigmentation and hypotrichosis following the lines of Blaschko, symmetric or asymmetric facial dysmorphism, and body asymmetry, in association with ocular, dental, and acral anomalies. Reported manifestations include microphthalmia, strabismus, myopia, oligodontia, microdontia, conical teeth, abnormal enamel, brachydactyly, syndactyly, and broad first toe, as well as dysmorphic facial features such as downslanting palpebral fissures, broad nasal bridge, malar hypoplasia, and microstomia. Brain imaging may show cystic leukoencephalopathy and ventricular dilation.

Dane

Klasyfikacja
Choroba

Synonimy
RHOA-related mosaic ectodermal dysplasia
Dysplazja ektodermalna mozaikowa związana z RHOA

Kod ORPHA
589608

Kod OMIM
-

Kod ICD10
Q82.4

Kod ICD11
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*Źródło

orphanet