

Opis choroby *

Definicja

Focal facial dermal dysplasia type III (FFDD3) is a rare focal facial dermal dysplasia (FFDD; see this term), characterized primarily by congenital bitemporal scar-like depressions and a typical, but variable facial dysmorphism, which may include distichiasis (upper lids) or lacking eyelashes, slanted eyebrows and a flattened and/or bulbous nasal tip and other features such as a low frontal hairline, sparse hair, redundant skin, epicanthal folds, low-set dysplastic ears, blepharitis and conjunctivitis.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

FFDD type III

FFDD type III

FFDD3

Ogniskowa dysplazja skóry twarzy 3, typ Setleisa

Zespół Setleisa

FFDD3

Focal facial dermal dysplasia 3, Setleis type

Setleis syndrome

Kod ORPHA

1807

Kod OMIM

227260

Kod ICD10

Q82.8

Kod ICD11

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*Źródło

orphanet