Opis choroby *

Definicja

A rare syndrome with intellectual disability, characterized by failure to thrive, short stature, joint laxity, soft skin, and distinctive facial features. Cardiac and neurological involvement is common and there is an increased lifetime risk of certain tumors. Costello syndrome belongs to the RASopathies, a group of conditions resulting from germline derived point mutations affecting the RAS-mitogen activated protein kinase pathway.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
FCS syndrome
Zespół FCS
Zespół twarzowo-skórno-szkieletowy
Faciocutaneoskeletal syndrome

Kod ORPHA
3071

Kod OMIM
218040

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet