

Opis choroby *

Definicja

Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome is characterised by the association of proximal fusion of the radius and ulna with congenital amegakaryocytic thrombocytopaenia. Less than 10 cases have been reported in the literature so far. The syndrome is transmitted as an autosomal dominant trait and is caused by mutations in the *HOXA11* gene (7p15).

Dane

Klasyfikacja

Zespół wad wrodzonych ATRUS syndrome
Zespół ATRUS

Synonimy

Kod ORPHA

71289

Kod OMIM

616738

Kod ICD10

Q87.2

Kod ICD11

3B64.01

*Źródło

orphanet