

## 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 thrombocytopenia. 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych ATRUS syndrome	ATRUS syndrome
	Zespół ATRUS

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
71289	616738	Q87.2

**Kod ICD11**  
3B64.01

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### \*Źródło

orphanet