

Opis choroby *

Definicja

A Noonan-related syndrome, characterized by facial anomalies suggestive of Noonan syndrome, loose anagen hair, frequent congenital heart defects, distinctive skin features (darkly pigmented skin, keratosis pilaris, eczema or ichthyosis), and short stature that is often associated with a growth hormone deficiency. Psychomotor delay with attention deficit/hyperactivity disorder (ADHD) is frequently observed.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Mazzanti syndrome
	NS/LAH
	Zespół Tosti
	NS/LAH

Kod ORPHA	Kod OMIM	Kod ICD10
2701	617506	Q87.1

Kod ICD11
LD2F.1Y

[*Źródło](#)

orphanet