

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

Zespół wad wrodzonych

Synonimy

Mazzanti syndrome

NS/LAH

Zespół Tosti

NS/LAH

Kod ORPHA

2701

Kod OMIM

617506

Kod ICD10

Q87.1

Kod ICD11

LD2F.1Y

[*Źródło](#)

orphanet