Opis choroby *

Definicja

Neurofibromatosis-Noonan syndrome (NFNS) is a RASopathy and a variant of neurofibromatosis type 1 (NF1) characterized by the combination of features of NF1, such as café-au-lait spots, iris Lisch nodules, axillary and inguinal freckling, optic nerve glioma and multiple neurofibromas, and Noonan syndrome (NS), such as short stature, typical facial features (hypertelorism, ptosis, downslanting palpebral fissures, low-set posteriorly rotated ears with a thickened helix, and a broad forehead), congenital heart defects and unusual pectus deformity. As these three entities have significant phenotypic overlap, molecular genetic testing is often necessary for a correct diagnosis (such as when café-au-lait spots are present in patients diagnosed with NS).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
NFNS
Nerwiakowłókniakowatość typu 1-zespół Noonan
NFNS
Neurofibromatosis type 1-Noonan syndrome

Kod ORPHA
638

Kod OMIM
601321

Kod ICD10
Q87.1

Kod ICD11
LD2F.1Y

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*Źródło

orphanet