Opis choroby *

Definicja

RIN2 syndrome, formerly known as macrocephaly, alopecia, cutis laxa and scoliosis (MACS) syndrome, is a very rare inherited connective tissue disorder characterized by macrocephaly, sparse scalp hair, soft-redundant and hyperextensible skin, joint hypermobility, and scoliosis. Patients have progressive facial coarsening with downslanted palpebral fissures, upper eyelid fullness/infraorbital folds, thick/everted vermillion, gingival overgrowth and abnormal position of the teeth. Rarer manifestations such as abnormal high-pitched voice, bronchiectasis, hypergonadotropic hypergonadism and brachydactyly (see this term) have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MACS syndrome
Niedobór RIN2
Zespół MACS
Zespół makrocefalii, łysienia, luźnej skóry i skoliozy
Macrocephaly-alopecia-cutis laxa-scoliosis syndrome
RIN2 deficiency
Tall forehead-sparse hair-skin hyperextensibility-scoliosis syndrome

Kod ORPHA
217335

Kod OMIM
613075

Kod ICD10
Q82.8

Kod ICD11
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orphanet