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 Synonimy

Zespół wad wrodzonych 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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*Źródło

orphanet