

DISEASE:
RIN2 syndrome

NAME:	RIN2 syndrome
DESCRIPTION:	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.
ORPHACODE:	217335
SYNONYMS:	MACS syndrome Macrocephaly-alopecia-cutis laxa-scoliosis syndrome RIN2 deficiency Tall forehead-sparse hair-skin hyperextensibility-scoliosis syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	RIN2
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