Opis choroby *

Definicja

A rare, genetic, premature aging disease characterized by sensorineural deafness, generalized lack of subcutaneous fatty tissue (although with increased truncal deposition) noted from childhood, scleroderma, and facial dysmorphism which includes prominent eyes, a beaked nose, small mouth, crowded teeth and mandibular hypoplasia. Other associated features include growth delay, joint contractures, telangiectasia, hypogonadism (with lack of breast development in females), cryptorchidism, skeletal muscle atrophy, hypertriglycemia and diabetes mellitus/insulin resistance.

Dane

Klasyfikacja
Choroba

Synonimy
MDP syndrome
Zespół MDP
MDPL syndrome
Mandibular hypoplasia-hearing loss-progeroid syndrome

Kod ORPHA
363649

Kod OMIM
615381

Kod ICD10
E34.8

Kod ICD11
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*Źródło

orphanet