Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome. ORPHA:363649

Mandibular hypoplasia-deafness-progeroid syndrome is 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.