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Progeroid and marfanoid aspect-lipodystrophy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Progeroid and marfanoid aspect-lipodystrophy syndrome. ORPHA:300382

Progeroid and marfanoid aspect-lipodystrophy syndrome is a rare systemic disease characterized by a neonatal progeroid appearance (not associated with other manifestations of premature aging) associated with facial dysmorphism (e.g. macrocephaly or arrested hydrocephaly, proptosis, downslanting palpebral fissures, retrognathia), generalized, extreme, congenital lack of subcutaneous fat tissue (except in the breast and iliac region) and incomplete signs of Marfan syndrome (mainly severe myopia, joint hyperextensibility and arachnodactyly). Metabolic disturbances are not associated.