Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome. ORPHA:137608

Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome is a rare, genetic, polymalformative syndrome characterized by progressive, proportionate, asymmetric segmental overgrowth (with soft tissue hypertrophy and ballooning effect) that develops and progresses rapidly in early childhood, arteriovenous and lymphatic vascular malformations, lipomatosis and linear epidermal nevus (arranged in whorls along the lines of Blaschko). Clinical symptoms of Cowden syndrome, such as macrocephaly and progressive development of numerous hypertrophic hamartomatous and neoplastic lesions involving multiple organs and systems, are also associated. Patients present an increased risk of developing cancer.