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Segmental progressive overgrowth syndrome with fibroadipose hyperplasia

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Segmental progressive overgrowth syndrome with fibroadipose hyperplasia. ORPHA:314662*

A rare PIK3CA-related overgrowth syndrome disease characterized by segmental and progressive overgrowth, predominantly involving the adipose tissue, or a mixture of adipose and fibrous tissue, with variable involvement of subcutaneous and muscular tissue, as well as skeletal overgrowth. Overgrowth severity and range is highly variable, although frequently it is asymmetric and disproportionate, it affects lower extremities more than the upper ones, and progresses in a distal to proximal pattern. Congenital overgrowth is typically associated.