Ferro-cerebro-cutaneous syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ferro-cerebro-cutaneous syndrome. ORPHA:397922

Ferro-cerebro-cutaneous syndrome is a rare, genetic, metabolic liver disease characterized by progressive neurodegeneration, cutaneous abnormalities, including varying degrees of ichthyosis or seborrheic dermatitis, and systemic iron overload. Patients manifest with infantile-onset seizures, encephalopathy, abnormal eye movements, axial hypotonia with peripheral hypertonia, brisk reflexes, cortical blindness and deafness, myoclonus and hepato/splenomegaly, as well as oral manifestations, including microdontia, wiedely spaced and pointed teeth with delayed eruption, and gingival overgrowth.