Bowen-Conradi syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Bowen-Conradi syndrome. ORPHA:1270

Bowen-Conradi syndrome (BCS) is a lethal autosomal recessive ribosomal biogenesis disorder characterized by severe prenatal and postnatal growth retardation, macrocephaly, a distinctive facial appearance, extreme psychomotor delay, hip and knee contractures and rockerbottom feet.