Epilepsy-microcephaly-skeletal dysplasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Epilepsy-microcephaly-skeletal dysplasia syndrome. ORPHA:1948

Epilepsy-microcephaly-skeletal dysplasia syndrome is characterized by the association of moderate to severe intellectual deficit, microcephaly, epilepsy, coarse face, hirsutism and skeletal abnormalities (scoliosis and retarded bone development). It has been described only once, in two sibs (one male and one female). This syndrome is likely to be an autosomal recessive condition and thus parents should be informed of a 25% risk of recurrence for other children.