Microcephaly-brachydactyly-kyphoscoliosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-brachydactyly-kyphoscoliosis syndrome. ORPHA:3433

Microcephaly-brachydactyly-kyphoscoliosis syndrome is characterized by profound intellectual deficit in association with microcephaly, short stature, brachydactyly type D, a flattened occiput, downslanting palpebral fissures, low-set large ears, a broad prominent nose and kyphoscoliosis. It has been described in three sisters. The disorder is likely to be transmitted as an autosomal recessive trait.