12q14 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 12q14 microdeletion syndrome. ORPHA:94063

12q14 microdeletion syndrome is characterised by mild intellectual deficit, failure to thrive, short stature and osteopoikilosis. It has been described in four unrelated patients. The syndrome appears to be caused by a heterozygous deletion at chromosome region 12q14, which was detected in three of the four patients. The deleted region contains the LEMD3 gene: mutations in this gene have already been implicated in osteopoikilosis.