Potocki-Shaffer syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Potocki-Shaffer syndrome. ORPHA:52022

Potocki-Shaffer syndrome is characterized by multiple exostoses, parietal foramina, enlargement of the anterior fontanelle and occasionally intellectual deficit and mild cranio-facial anomalies. To date, 23 individuals from 14 families have been reported. The syndrome is caused by contiguous gene deletions on the short arm of chromosome 11 (11p11.2).