Seizures-scoliosis-macrocephaly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Seizures-scoliosis-macrocephaly syndrome. ORPHA:466926

Seizures-scoliosis-macrocephaly syndrome is a rare, genetic neurometabolic disorder characterized by seizures, macrocephaly, delayed motor milestones, moderate intellectual disability, scoliosis with no exostoses, muscular hypotonia present since birth, as well as renal dysfunction. Coarse facial features (including hypertelorism and long hypoplastic philtrum) and bilateral cryptorchidism (in males) are also commonly reported. Additional manifestations include abnormal gastrointestinal motility (resulting in constipation, diarrhea, gastroesophageal reflux and dysphagia), gait disturbances, strabismus and ventricular septal defects.