Open Peer Review on Qeios

Macrocephaly-intellectual disabilityautism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Macrocephaly-intellectual disability-autism syndrome</u>. ORPHA:210548

A rare, genetic, neurological disease characterized by association of macrocephaly, dysmorphic facial features and psychomotor delay leading to intellectual disability and autism spectrum disorder. Facial dysmorphism may include frontal bossing, hypertelorism, midface hypoplasia, depressed nasal bridge, short nose, and long philtrum.