THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome.
ORPHA:363444

THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by global development delay, microcephaly, moderate to severe intellectual disability and facial dysmorphism which includes tall forehead, high anterior hairline, short upslanting palpebral fissures, deep-set eyes and a long nose with a low-hanging columella. Additionally, congenital renal and cardiac malformations (such as horseshoe kidney, unilateral renal agenesis atrioventricular septal defects, patent ductus arteriosus), as well as corpus callosum dysplasia, may be associated.