Open Peer Review on Qeios

PDE4D haploinsufficiency syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. PDE4D haploinsufficiency syndrome. ORPHA:439822

PDE4D haploinsufficiency syndrome is a rare syndromic intellectual disability characterized by developmental delay, intellectual disability, low body mass index, long arms, fingers and toes, prominent nose and small chin.