Intellectual disability, Buenos-Aires type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Intellectual disability, Buenos-Aires type. ORPHA:3079

Intellectual disability, Buenos-Aires type is a rare intellectual disability syndrome characterized by growth retardation, microcephaly, characteristic facial features (including narrow forehead, bushy eyebrows, hypertelorism, small, downward-slanting palpebral fissures with blepharoptosis, malformed and low-set ears, broad straight nose, thin upper lip, and a wide, tented mouth), developmental delay, intellectual disability, speech disorder, and multiple organ malformations (e.g. ventricular septal defect, megaloureter, dilated renal pelvis). Additional manifestations reported include neurocutaneous lesions (including palmoplantar hyperkeratosis), internal hydrocephalus, and bilateral partial soft-tissue syndactyly of second and third toe.