PACS1 Related Syndrome

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We report on 19 individuals with a recurrent de novo c.607C>T mutation in PACS1. This specific mutation gives rise to a recognizable intellectual disability syndrome. There is a distinctive facial appearance (19/19), characterized by full and arched eyebrows, hypertelorism with downslanting palpebral fissures, long eye lashes, ptosis, low set and simple ears, bulbous nasal tip, wide mouth with downturned corners and a thin upper lip with an unusual "wavy" profile, flat philtrum, and diastema of the teeth. Intellectual disability, ranging from mild to moderate, was present in all. Hypotonia is common in infancy (8/19). Seizures are frequent (12/19) and respond well to anticonvulsive medication. Structural malformations are common, including heart (10/19), brain (12/16), eye (10/19), kidney (3/19), and cryptorchidism (6/12 males). Feeding dysfunction is presenting in infancy with failure to thrive (5/19), gastroesophageal reflux (6/19), and gastrostomy tube placement (4/19). There is persistence of oral motor dysfunction. We provide suggestions for clinical work-up and management and hope that the present study will facilitate clinical recognition of further cases.