Acrocephalopolydactyly

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acrocephalopolydactyly. ORPHA:221054

Acrocephalopolydactyly, also known as Elejalde syndrome, is an extremely rare lethal autosomal recessive disorder characterized by massive birth weight, swollen globular body, generalized edema, short limbs, postaxial polydactyly, thick skin, facial dysmorphism (slanted palpebral fissures, hypertelorism, epicanthic folds, dysplastic ears), excessive connective tissue, renal dysplasia, and in some patients, organomegaly, craniosynostosis with acrocephaly, omphalocele, cleft palate, and cryptorchidism. Fewer than 10 cases have been reported to date.