Arthrogryposis multiplex congenita-whistling face syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Arthrogryposis multiplex congenita-whistling face syndrome. ORPHA:1150

Arthrogryposis multiplex congenita-whistling face syndrome is an extremely rare type of arthrogryposis multiplex congenita characterized by the combination of multiple joint contractures with movement limitation, microstomia with a whistling appearance of the mouth that may cause feeding, swallowing, and speech difficulties, a distinctive expressionless facies, severe developmental delay, central and autonomous nervous system dysfunction (excessive salivation, temperature instability, myoclonic epileptic fits, bradycardia), occasionally Pierre-Robin sequence, and lethality generally occurring during the first months of life. Arthrogryposis multiplex congenita-whistling face syndrome has been suggested to be a fetal akinesia deformation sequence.