3MC syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 3MC syndrome. ORPHA:293843

3MC syndrome describes a rare developmental disorder, that unifies the overlapping autosomal recessive disorders previously known as Carnevale, Mingarelli, Malpuech and Michels syndromes, characterized by a spectrum of developmental anomalies that include distinctive facial dysmorphism (i.e. hypertelorism, blepharophimosis, blepharoptosis, highly arched eyebrows), cleft lip and/or palate, craniosynostosis, learning disability, radioulnar synostosis and genital and vesicorenal anomalies. Less common features reported include anterior chamber defects, cardiac anomalies (e.g. ventricular septal defect; see this term), caudal appendage, umbilical hernia/omphalocele and diastasis recti.