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Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7. ORPHA:96182*

Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7 is a genetic malformation syndrome with short stature characterized by severe prenatal and postnatal growth retardation, feeding difficulties, body asymmetry, dysmorphic craniofacial features (triangular-shaped face, relative macrocephaly, frontal bossing, micrognathia, down-turned corners of the mouth) and other anomalies (fifth finger clinodactyly, café au lait macules, male genital anomalies, mild developmental delay and/or speech delay with movement disorders).