Maternal uniparental disomy of chromosome 16

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Maternal uniparental disomy of chromosome 16. ORPHA:96185

Maternal uniparental disomy of chromosome 16 is a uniparental disomy of maternal origin which might be associated with intrauterine growth retardation and an elevated risk of congenital malformations. Healthy carriers have also been reported. In addition, cases of homozygosity for a recessive disease mutation for which the mother was a carrier have been described, and specific phenotype depends on the inherited disorder.