Open Peer Review on Qeios

Trisomy 8q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Trisomy 8q. ORPHA:1752

A partial autosomal trisomy characterized by developmental delay, intellectual disability, prenatal and postnatal growth retardation, congenital heart, genitourinary and skeletal anomalies, and dysmorphic facial features, including high and broad forehead, hypertelorism, upslanting palpebral fissures, broad nose, dysplastic and low set ears, micrognathia. Phenotypic features vary in relation to the duplication size.