# 7p22.1 microduplication syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 7p22.1 microduplication syndrome. ORPHA:314034*

7p22.1 microduplication syndrome is a rare chromosomal anomaly syndrome, resulting from a partial interstitial microduplication of the short arm of chromosome 7, characterized by intellectual disability, psychomotor and speech delays, craniofacial dysmorphism (including macrocephaly, frontal bossing, hypertelorism, abnormally slanted palpebral fissures, anteverted nares, low-set ears, microretrognathia) and cryptorchidia. Cardiac (e.g., patent foramen ovale and atrial septal defect), as well as renal, skeletal and ocular abnormalities may also be associated.