8p inverted duplication/deletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 8p inverted duplication/deletion syndrome. ORPHA:96092

8p inverted duplication/deletion [invdupdel(8p)] syndrome is a rare chromosomal anomaly characterized clinically by mild to severe intellectual deficit, severe developmental delay (psychomotor and speech development), hypotonia with tendency to develop progressive hypertonia and severe orthopedic problems over time, minor facial anomalies and agenesis of the corpus callosum.