3q26q27 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 3q26q27 microdeletion syndrome. ORPHA:356947

3q26q27 microdeletion syndrome is a rare partial autosomal monosomy syndrome characterized by neonatal hypotonia, prenatal and postnatal growth deficiency, severe feeding difficulties, global developmental delay and intellectual disability, dental anomalies (delayed tooth eruption, delayed loss of primary teeth, dental crowding), recurrent respiratory infections, thrombocytopenia and facial dysmorphism (flat facial profile, medially sparse eyebrows, epicanthal folds, flat nasal bridge and tip, short philtrum). Behavioral abnormalities (ADHD, Asperger syndrome) have also been reported.