2p21 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 2p21 microdeletion syndrome. ORPHA:163693

The 2p21 microdeletion syndrome consists of cystinuria, neonatal seizures, hypotonia, severe growthand developmental delay, facial dysmorphism, and lactic acidemia.