2q31.1 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 2q31.1 microdeletion syndrome. ORPHA:251014

2q31.1 microdeletion syndrome is a well-defined and clinically recognisable syndrome characterized by moderate to severe developmental delay, short stature, facial dysmorphism and variable limb defects.