Stimmler syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Stimmler syndrome. ORPHA:3199

Stimmler syndrome is characterised by the association of microcephaly, low birth weight and severe intellectual deficit with dwarfism, small teeth and diabetes mellitus. Two cases have been described. Biochemical tests reveal the presence of high levels of alanine in the urine and elevated alanine, pyruvate and lactate levels in the blood.