Hernández-Aguirre Negrete syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hernández-Aguirre Negrete syndrome. ORPHA:2139

Hernández-Aguirre Negrete syndrome is characterized by major seizures, dysmorphic features (round face, bulbous nose, wide mouth, prominent philtrum), pes planus, psychomotor retardation and obesity. It has been described in five children (three boys and two girls, one of whom died in infancy) from two unrelated Mexican families. This condition is likely to be transmitted as an autosomal recessive trait.