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Pili torti-developmental delay-neurological abnormalities syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pili torti-developmental delay-neurological abnormalities syndrome. ORPHA:2891

Pili torti-developmental delay-neurological abnormalities syndrome is characterized by growth and developmental delay, mild to moderate neurologic abnormalities, and pili torti. It has been described in a brother and his sister born to consanguineous Puerto Rican parents.