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Autosomal recessive infantile hypercalcemia

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Autosomal recessive infantile hypercalcemia. ORPHA:300547*

Autosomal recessive infantile hypercalcemia is a rare, genetic, phosphocalcic metabolism disorder characterized by early-onset hypercalcemia, hypophosphatemia, hypercalciuria, decreased intact parathyroid hormone serum levels and medullary nephrocalcinosis, typically manifesting with failure to thrive, hypotonia, vomiting, constipation and/or polyuria.