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Central nervous system calcification-deafness-tubular acidosis-anemia syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Central nervous system calcification-deafness-tubular acidosis-anemia syndrome. ORPHA:3240*

This syndrome is characterised by progressive calcification of the brain and spinal cord, growth retardation, psychomotor anomalies, deafness and anaemia. Renal tubular acidosis was found in one patient. To date, this syndrome has been described in only two patients from one family.