AICA-ribosiduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. AICA-ribosiduria. ORPHA:250977

AICA-ribosiduria is an extremely severe inborn error of purine biosynthesis characterized clinically in the single reported case to date by profound intellectual deficit, epilepsy, dysmorphic features of the knees, elbows, and shoulders and congenital blindness.