Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome. ORPHA:314572

Autosomal recessive leukoencephalopathy-ischemic stroke-retinitis pigmentosa syndrome is a rare neurologic disease characterized by global developmental delay, intellectual disability, multiple ischemic lesions in brain MRI, behavioral abnormalities, dystonia, choreic movements and pyramidal syndrome, facial dysmorphism (hypertelorism, arched palate, macroglossia), retinitis pigmentosa, scoliosis, seizures.