Leigh syndrome with nephrotic syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Leigh syndrome with nephrotic syndrome. ORPHA:255249

A rare, genetic neurometabolic disease characterized by encephalomyopathy (including developmental delay, nystagmus, progressive ataxia, dystonia, amyotrophy, visual loss, sensorineural deafness, seizures) and bilateral, symmetrical lesions in the basal ganglia or brainstem on imaging, associated with nephrotic syndrome.