Microphthalmia-brain atrophy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microphthalmia-brain atrophy syndrome. ORPHA:77299

Microphthalmia-brain atrophy (MOBA) syndrome is a rare genetic neurodegenerative disorder characterized by congenital microphthalmia, sunken eyes, blindness, microcephaly, severe intellectual disability, progressive spasticity, and seizures. Psychomotor development is normal in the first 6-8 months of life and thereafter declines rapidly and continuously. Brain MRI reveals progressive and extensive degenerative changes, especially cortex, cerebellum, brainstem, and corpus callosum atrophy, with complete loss of cerebral white matter.