Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome. ORPHA:352654

Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome is a genetic neurodegenerative disease characterized by normal early development followed by childhood onset optic atrophy with progressive vision loss and eventually blindness, followed by progressive neurological decline that typically includes cerebellar ataxia, nystagmus, dorsal column dysfunction (decreased vibration and position sense), spastic paraplegia and finally tetraparesis.