

Kearns-Sayre Syndrome

Kearns-Sayre syndrome is a mitochondrial disease characterized by ophthalmoplegia, retinitis pigmentosa, and cardiac electrophysiology like heart block.



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Characteristics

Mitochondrial Disease

Damaged Mitochondria

Kearns-Sayre syndrome is a multi-organ disorder caused by a defect in mitochondrial genes. In rare cases, it can be autosomal dominant as well.

Clinical Findings

Ophthalmoplegia

Ophthalmoscope-crazy-eye

Chronic progressive external ophthalmoplegia (CPEO) is present in patients with this disease. It is a type of eye disorder characterized by a slowly progressive inability to move the eyes and eyebrows.

Retinitis Pigmentosa

Red-tin-eyes Piggy-bank

Retinitis pigmentosa is a hereditary degenerative disease of the retina which involves excessive pigmentation and visual field defects.

Heart Block

Heart Blocks

Heart block occurs due to disruptions in the heart's pacemaker current that passes from the atria to the ventricles. Patients present with bradycardia and may have characteristic EKG findings.