CHAPTER 30

Mitochondrial Disorder: Kearns-Sayre Syndrome

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General Features

- Mitochondrial diseases are multisystem disorders: anemia, myopathy, lactic acidosis, CNS abnormality, endocrine abnormalities, renal disease, sensorineural deafness, and retinal involvement. The clinical abnormalities are heterogeneous, and they usually begin in childhood. Premature death occurs because of cardiac conduction defects.
- The onset is usually before 20 years of age. The fundus shows pigmentary retinopathy, with a salt-and-pepper appearance (Fig. 30.1), but vision remains good in most patients.
- Systemic involvement includes chronic progressive external ophthalmoplegia (CPEO), with ptosis being the most common complaint, and cardiomyopathy.
- Other variable features are short stature; cerebellar symptoms; weakness of muscles of the face, pharynx, trunk, or extremities; and progressive hearing loss.
- Full-field ERG does show evidence of generalized retinal dysfunction, involving both rods and cones.
- Skeletal muscle biopsy shows ragged red fibers and abnormal mitochondria.

Molecular Genetics

Mitochondrial DNA (mtDNA) deletions can be identified, and rarely point mutation.

Suggested Reading

Ortiz A, Arias J, Cárdenas P, Villamil J, Peralta M, Escaf LC, Ortiz J. Macular findings in spectral domain optical coherence tomography and OCT angiography in a patient with Kearns-Sayre syndrome. Int J Retina Vitreous. 2017;3:24.

Shemesh A, Margolin E. Kearns Sayre syndrome. Treasure Island: StatPearls Publishing; 2018.

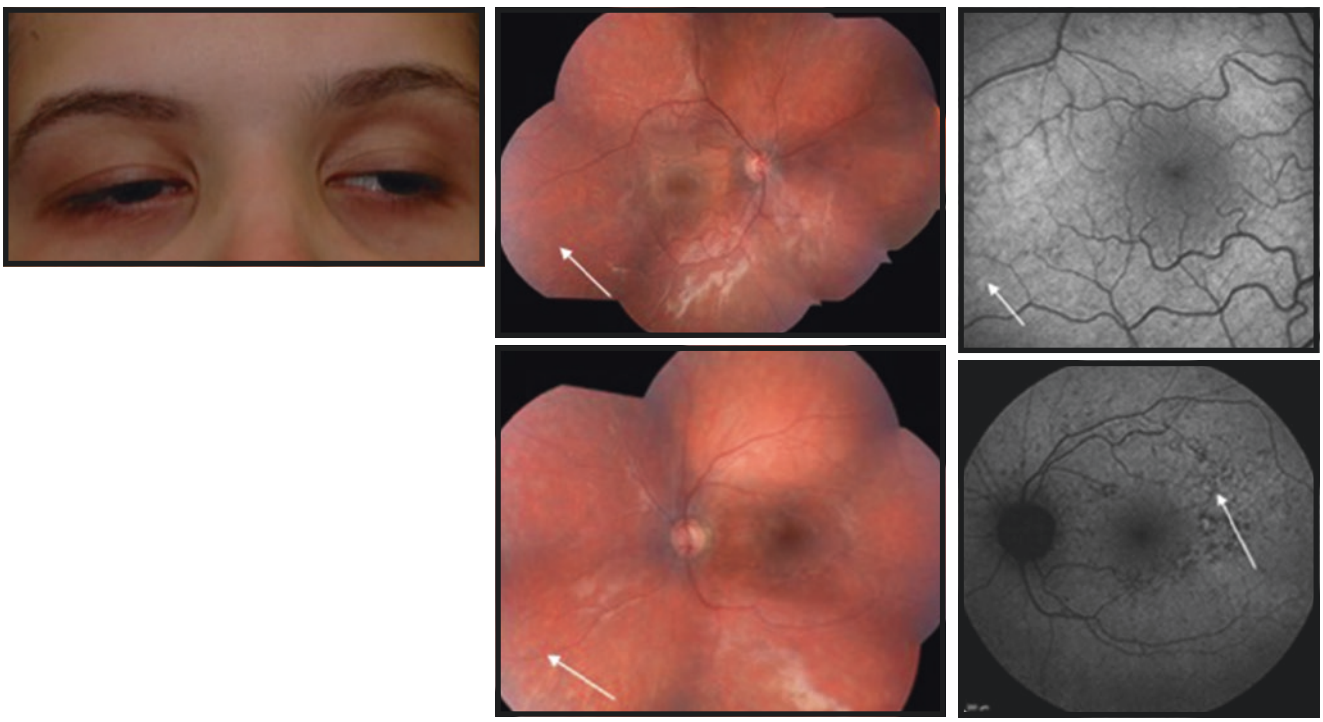

Fig. 30.1 Kearns-Sayre syndrome in this patient is evidenced by ptosis, more marked in the right eye; pigmentary retinopathy (*arrows*), and a stippled autofluorescence pattern (*arrows*)