

Kearns-Sayre syndrome (KSS) is a rare neuromuscular disorder. An important clinical symptomatic feature is the presence of a mono-or bilateral ptosis (partial closure of the eyelids). This disease is mostly characterized by three primary findings: progressive paralysis of certain eye muscles (chronic progressive external ophthalmoplegia [CPEO]); abnormal accumulation of colored (pigmented) material on the nerve-rich membrane lining the eyes (atypical retinitis pigmentosa), leading to chronic inflammation, progressive degeneration, and wearing-away of certain eye structures (pigmentary degeneration of the retina); and heart disease (cardiomyopathy) such as heart block. Other findings may include muscle weakness, short stature, hearing loss, and/or the loss of ability to coordinate voluntary movements (ataxia) due to problems affecting part of the brain (cerebellum). In some cases, KSS may be associated with other disorders and/or conditions.