Supplementary Table 1. Genotypic and phenotypic summary of reported cases of primary coenzyme Q10 deficiency with ocular manifestations.

Reference	Genotype	Phenotype – Ocular	Phenotype – Systemic
CoQ2			
Abdelhakim et al.[7]	*CoQ2* c.[288dupC];[376C > G] p.[(Ala97Argfs*56)];[(Arg126Gly)]	Sibling 1: retinopathy* Sibling 2: retinopathy, optic atrophy, posterior subcapsular cataract Sibling 3: retinopathy, posterior subcapsular cataract	Nephrotic syndrome
Mitsui et al.[24] Hara et al.[5]	*CoQ2* c.[382A>G] p.[(Met128Val)]	Two siblings; retinopathy	Multiple-system atrophy
Salviati et al.[6] Quinzii et al.[18] Diomedi-Camassei et al.[9]	*CoQ2* c.890A>G p.[Tyr297Cys)]	Retinopathy, optic atrophy	Nephrotic syndrome, hypotony, encephalopathy
PDSS1/2			
Nardecchia et al.[2]	*PDSS1* c.735G>T p.Gln245His	Two siblings; optic atrophy	Sensorineural hearing loss, cardiac valvulopathy, mild intellectual disability
Mollet et al.[3]	*PDSS1* c.977T>G p.Asp308Glu	Two siblings; optic atrophy	Sensorineural hearing loss, cardiac valvulopathy, mild intellectual disability, peripheral neuropathy
Other			
Rotig et al.[4]	Unknown (molecular diagnosis)	Sibling 1: retinopathy optic atrophy, cataract Sibling 2: unspecified vision loss	Nephrotic syndrome, ataxia, dystonia

Boitier et al.[19]	Unknown (molecular diagnosis)	Retinopathy	Cerebellar ataxia, epilepsy

*“Retinopathy” encompasses the broad range of phenotypes associated with CoQ10 deficiency described in the literature, including rod-cone dystrophy, pigmentary retinopathy, and retinitis pigmentosa-like disease.