Mutations in DNA Polymerase γ (POLG)

Human POLG Gene

Mitochondrial Targeting Sequence

Exonuclease Domain

Linker Region

Polymerase Domain

Progressive External Ophthalmoplegia (PEO) [adPEO] Sporadic Alpers, Myocerebrohepatopathy and other Infantile Hepatocerebral Syndromes with mtDNA depletion

Ataxia-Neuropathy Syndrome, MIRAS, SANDO, SCAE, Friedreich’s Ataxia

Male Infertility, Testicular Cancer, Idiopathic Parkinsons [NRTI Toxicity]

Charcot-Marie Tooth Disease [Leigh’s Syndrome] [Other or Unclear]

Single Nucleotide Polymorphism ↓