



THYMIDINE KINASE 2 DEFICIENCY:

From Diagnostic Odyssey to Timely Diagnosis and Treatment

Clinical Resources

Clinical Trials (<https://www.clinicaltrials.gov>)

NCT03639701 – Treatment of TK2 Deficiency With Thymidine and Deoxycytidine

NCT03845712 – An Open-Label Study of Continuation Treatment With Combination Pyrimidine Nucleosides in Patients With TK2 Deficiency

Guidelines

Parikh S, Goldstein A, Karaa A, et al. Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Genet Med.* 2017;19(12):1-18. doi:10.1038/gim.2017.107

Mavraki E, Labrum R, Sergeant K, et al. Genetic testing for mitochondrial disease: the United Kingdom best practice guidelines. *Eur J Hum Genet.* 2023;31:148-163. doi:10.1038/s41431-022-01249-w

Other Resources

- Medline Plus: [TK2-related mitochondrial DNA depletion syndrome, myopathic form](#)
- Mitochondrial Care Network: [What is the Mitochondrial Care Network \(MCN\)?](#)
- Mitochondrial Disease Action Committee (MitoAction): [Mitochondrial Disease](#)
- National Organization for Rare Diseases: [Thymidine Kinase 2 Deficiency](#)
- The Mitochondrial Medicine Society: [Mito U Clinician Resource Library](#)
- TK2d: [What is TK2d?](#)
- United Mitochondrial Disease Foundation: [Understanding and Navigating Mitochondrial Disease](#)