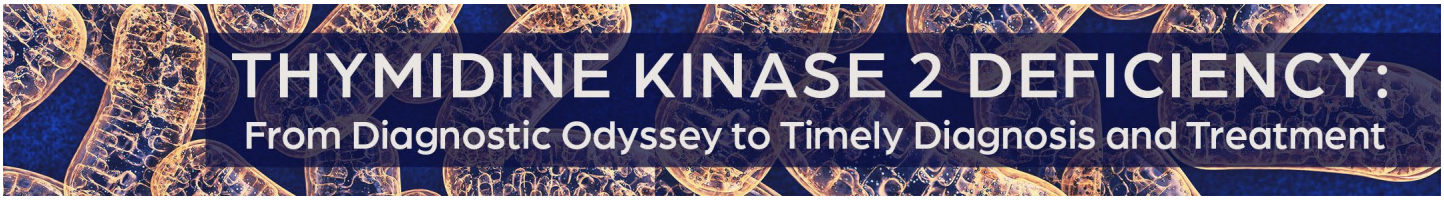

Reference List

Andrieux P, Chevillard, C, Cunha-Neto E, Nunes JPS. Mitochondria as a cellular hub in infection and inflammation. *Int J Mol Sci.* 021;22:11338. doi:10.3390/ijms222111338

Berardo A, Dominguez-Gonzalez C, Engelstad K, Hirano M. Advances in thymidine kinase 2 deficiency: clinical aspects, translational progress, and emerging therapies. *J Neuromusc Dis.* 2022;9:225-235. doi:10.3233/JND-210786

Zogenix MDS, Inc. A study of the efficacy and safety of MT1621 in thymidine kinase 2 (TK2) deficiency (treatment naïve). Updated September 1, 2023. Accessed October 20, 2023. https://clinicaltrials.gov/study/NCT04581733?intr=MT1621&rank=1

Garone C, Taylor RW, Nascimento A, et al. Retrospective natural history of thymidine kinase 2 deficiency. *J Med Genet.* 2018;55:515-521. doi:10.1136/jmedgenet-2017-105012

Grier J, Hirano M, Karaa A, Shepard E, Thompson JLP. Diagnostic odyssey of patients with mitochondrial disease: results of a survey. *Neurol Genet.* 2018;4(2):e230. doi:10.1212/NXG.0000000000000230

Khan NA, Govindaraj P, Meena AK, Thangaraj K. Mitochondrial disorders: challenges in diagnosis and treatment. *Indian J Med Res.* 2015;141:13-26. doi:10.4103/0971-5916.154489

Klopstock T, Priglinger C, Yilmaz A, et al. Mitochondrial disorders. *Dtsch Arztebl Int.* 2021;118: 741–748. doi:10.3238/arztebl.m2021.0251

Lopez-Gomez C, Levy RJ, Sanchez-Quintero MJ, et al. Deoxycytidine and deoxythymidine treatment for thymidine kinase 2 deficiency. *Ann Neurol.* 2017;81:641-652. doi:10.1002/ana.24922

Mitoaction.org. Mitochondrial Care Network. Published 2023. Accessed October 20, 2023. https://www.mitoaction.org/mitochondrial-disease/doctors/mcn/

Muraresku CC, McCormick EM, Falk MJ. Mitochondrial disease: advances in clinical diagnosis, management, therapeutic development, and preventative strategies. *Curr Genet Med Rep.* 2018;6:62-72. doi:10.1007/s40142-018-0138-9

National Organization for Rare Disorders (NORD). Thymidine kinase 2 deficiency. Updated September 14, 2022. Accessed October 20, 2023. https://rarediseases.org/rare-diseases/thymidine-kinase-2-deficiency/

O'Ferrall E. Mitochondrial disorders: treatment. UpToDate. Updated November 22, 2022. Accessed October 20, 2023. https://www.uptodate.com/contents/mitochondrial-disorders-treatment

Parikh S, Goldstein A, Koenig MK, et al. Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Genet Med.* 2015;17(9):689-701. doi:10.1038/gim.2014.177

Quan J, Domínguez-González C, Paradas C, et al. A RETROspective Study of Combination Pyrimidine Nucleoside Therapy in Patients with Thymidine Kinase 2 (TK2) Deficiency. Presented at: World Muscle Society Annual Meeting; October 1-5, 2019; Copenhagen, Denmark.

Raymond FL, Horvath R, Chinnery PF. First-line genomic diagnosis of mitochondrial disorders. *Nat Rev Genet.* 2018;19:399-400.
doi:10.1038/s41576-018-0022-1

Takeontk2d. Published 2023. Accessed October 20, 2023. https://www.tk2d.com/#managed

TheLilyFoundation.org. Mito research around the world. Published 2022. Accessed October 20, 2023. https://www.thelilyfoundation.org.uk/lily-research/what-research-going-world/151/

United Mitochondrial Disease Foundation. Published 2023. Accessed October 20, 2023. https://www.umdf.org/research/mitochondrial-genetic-testing/

Watson E, Davis R, Sue CM. New diagnostic pathways for mitochondrial disease. *J Transl Genet Genom.* 2020;4:188-202.
doi:10.20517/jtgg.2020.31

Wei W, Chinnery PF. Inheritance of mitochondrial DNA in humans: implications for rare and common diseases. *J Intern Med.* 2020;287:634-644.
doi:10.1111/joim.13047