

## References

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1. Hirano M. (Updated January 2016). Mitochondrial Neurogastrointestinal Encephalopathy Disease. In: GeneReviews at GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2017. Available at: [www.ncbi.nlm.nih.gov/books/NBK1179/](http://www.ncbi.nlm.nih.gov/books/NBK1179/).
2. De Giorgio R, Pironi L, Rinaldi R, et al. Liver transplantation for mitochondrial neurogastrointestinal Encephalomyopathy. *Annals of Neurology*. 2016;80(3):448–455.
3. Halter J, Schüpbach W, Casali C, et al. Allogeneic hematopoietic SCT as treatment option for patients with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): A consensus conference proposal for a standardized approach. 2010;46(3).
4. Sivadasan A, Muthusamy K, Patil AK, et al. (2016) Pearls & Oysters: Mitochondrial neurogastrointestinal Encephalomyopathy. *Neurology*. 2016;86(14):147–150.
5. OMIM entry - \* 131222 - THYMIDINE PHOSPHORYLASE; TYMP (1992) Available at: [omim.org/entry/131222?search=tymp&highlight=tymp](http://omim.org/entry/131222?search=tymp&highlight=tymp).
6. Parikh S, Goldstein A, Koenig MK, et al. Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. *Gen in Med*. 2015;(17):689-701.
7. Finsterer J, Harbo HF, Baets J, et al. EFNS guidelines on the molecular diagnosis of mitochondrial disorders. *Eur J Neurol*. 2009;16(12):1255-64.
8. Nesbitt, V, Alston CL, Blakely EL, et al. A national perspective on prenatal testing for mitochondrial disease. *Eur J Hum Genet*. 2014;22(11).
9. Nishino I, Spinazzola A, Hirano M. Thymidine phosphorylase gene mutations in MNGIE, a human mitochondrial disorder. *Science*. 1999;283:689–92.
10. Garone C, Tadesse S, Hirano M. Clinical and genetic spectrum of mitochondrial neurogastrointestinal encephalomyopathy. *Brain*. 2011;134:3326–32.