North American Mitochondrial Diseases Consortium

Journal Articles

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3. Marti R, Dorado B, Hirano M. Measurement of mitochondrial dNTP pools. *Methods Mol. Biol.* 2012;837:135-148. PMID: 22215545
4. Marti R, Lopez LC, Hirano M. Assessment of thymidine phosphorylase function: measurement of plasma thymidine (and deoxyuridine) and thymidine phosphorylase activity. *Methods Mol. Biol.* 2012;837:121-133. PMID: 22215544
5. Rahman S, Clarke CF, Hirano M. 176th ENMC International Workshop: diagnosis and treatment of coenzyme Q(1)(0) deficiency. *Neuromuscul. Disord.* Jan 2012;22(1):76-86. PMID: 21723727, PMCID: PMC3222743
6. Pascual JM, Liu P, Mao D, et al. Triheptanoin for glucose transporter type I deficiency (G1D): modulation of human ictogenesis, cerebral metabolic rate, and cognitive indices by a food supplement. *JAMA neurology.* Oct 2014;71(10):1255-1265. PMID: 25110966
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10. Navarro-Gomez D, Leipzig J, Shen L, et al. Phy-Mer: a novel alignment-free and reference-independent mitochondrial haplogroup classifier. *Bioinformatics.* Apr 15 2015;31(8):1310-1312. PMID: 25505086, PMCID: PMC4393525
11. Al-Mehmadi S, Splitt M, Ramesh V, et al. FHF1 (FGF12) epileptic encephalopathy. *Neurology Genetics.* 2016;2(6):e115. PMID: 27830185, PMCID: PMC5087254
12. Servian-Morilla E, Takeuchi H, Lee TV, et al. A POGLUT1 mutation causes a muscular dystrophy with reduced Notch signaling and satellite cell loss. *EMBO Mol Med.* 2016;8(11):1289-1309. PMID: 27807076, PMCID: PMC5090660
13. Marin SE, Saneto RP. Neuropsychiatric Features in Primary Mitochondrial Disease. Neurol. Clin. Feb 2016;34(1):247-294. PMID: 26614002