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EXPERIENCES AND POSITION :

- ◆ Professor of Neurology and Pathology & Cell Biology Neuromuscular Disease

EDUCATIONAL BACKGROUND :

- ◆ Harvard College, A.B.
- ◆ Albert Einstein College of Medicine, M.D.
- ◆ Neurology Residency, Neurological Institute of New York, The New York-Presbyterian Hospital at Columbia University Medical Center
- ◆ Neuromuscular Disease Fellowship, Neurological Institute of New York, The New York-Presbyterian Hospital at Columbia University Medical Center

RESEARCH INTERESTS :

I have been studying genetic neuromuscular diseases with a focus on mitochondrial diseases. My laboratory has identified causative mutations for several diseases including: mitochondrial neurogastrointestinal encephalomyopathy (MNGIE), LAMP2 deficiency, X-linked scapuloperonal myopathy, Navajo neurohepatopathy, and coenzyme Q10 deficiencies. We are studying cell and mouse models of MNGIE, coenzyme Q10 deficiency, and mitochondrial DNA depletion due to thymidine kinase 2 (TK2) deficiency. We are also testing therapies for MNGIE. Together with Dr. Salvatore DiMauro, Dr. Hirano is developing a North American Mitochondrial Disease Consortium (NAMDC).

PUBLISHED WORKS : (Selected List)

- Hirano M, Silvestri G, Blake DM, Lombes A, Minetti C, Bonilla E, Hays AP, Lovelace RE, Butler I, Bertorini TE, Threlkeld AB, Mitsumoto H, Salberg L, Rowland LP, DiMauro S. (1994) Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): clinical, biochemical, and genetic features of an autosomal recessive mitochondrial disorder. *Neurology*. 44:721-727
- Hirano M, Yebenes J, Jones AC, Nishino I, DiMauro S, Carlo JR, Bender AN, Hahn AF, Salberg LM, Weeks DE, Nygaard TG (1998) Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE) syndrome maps to chromosome 22q13.32-qter. *Am J Hum Genet* 63:526-533
- Nishino I, Spinazzola A, Hirano M. (1999) Thymidine phosphorylase gene mutations in MNGIE, a human mitochondrial disorder. *Science* 283:689-692
- Nishino I, Fu J, Tanji K, Yamada T, Shimojo S, Koori T, Mora M, Riggs JE, Oh SJ, Koga Y, Sue CM, Yamamoto A, Morakami N, Shanske S, Byrne E, Bonilla E, Nonaka I, DiMauro S, Hirano M (2000) Primary LAMP-2 deficiency causes X-linked vacuolar cardiomyopathy and myopathy (Danon Disease). *Nature* 406:906-910
- Spinazzola A, Marti R, Nishino I, Andreu A, Naini A, Tadesse S, Pela I, Zammarchi E, Donati MA, Oliver JA, Hirano M (2002) Altered thymidine metabolism due to defects of thymidine phosphorylase. *J Biol Chem* 277:4128-4133 Nishigaki Y, Martí R, Copeland WC, Hirano M (2003) Site-specific mtDNA point mutations due to thymidine phosphorylase deficiency. *J Clin Invest* 111:1913-1921
- Quinzii C, Naini A, Salviati L, Trevisson E, Navas P, DiMauro S, Hirano M (2006) A Mutation in Para-Hydroxybenzoate-Polyprenyl Transferase (COQ2) Causes Primary Coenzyme Q10 Deficiency. *Am J Hum Genet* 78:345-349
- López LC, Schuelke M, Quinzii C, Kanki T, Rodenburg RJT, Hirano M (2006) Leigh syndrome with nephropathy and CoQ10 deficiency due to *decaprenyl diphosphate synthase subunit 2 (PDSS2)* mutations. *Am J Hum Genet* 79:1125-1129
- Hirano M, Marti R, Casali C, Tadesse S, Uldrick T, Fine B, Escolar DM, Valentino ML, Nishino I, Hesdorffer C, Schwartz J, Hawks RG, Martone DL, Cairo MS, DiMauro S, Stanzani M, Garvin JH, Jr., Savage DG (2006) Allogeneic stem cell transplantation corrects biochemical derangements in MNGIE. *Neurology* 67:1458-1460
- Akman HO, Dorado B, Lopez LC, Garcia-Cazorla A, Vila MR, Tanabe LM, Dauer WT, Bonilla E, Tanji K, Hirano M (2008) Thymidine kinase 2 (H126N) knockin mice show the essential role of balanced deoxynucleotide pools for mitochondrial DNA maintenance. *Hum Mol Genet* 17:2433-2440
- Lopez LC, Akman HO, Garcia-Cazorla A, Dorado B, Marti R, Nishino I, Tadesse S, Pizzorno G, Shungu D, Bonilla E, Tanji K, Hirano M (2009) Unbalanced deoxynucleotide pools cause mitochondrial DNA instability in thymidine phosphorylase deficient mice. *Hum Mol Genet* 18:714-722