



The 6th Congress of Asian Society for Pediatric Research & 51st Annual Meeting of Taiwan Pediatric Association

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CURRENT POSITION :

- ◆ Director, Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry
- ◆ Guest Professor, Waseda University Faculty of Science and Engineering

EDUCATIONAL BACKGROUND :

- ◆ 1983-1989 Kyoto University Medical School
- ◆ 1989-1992 Neurology Residency, Kyoto University Hospital and affiliated hospitals
- ◆ 1994-1998 Neuromuscular Research Fellow, National Institute of Neuroscience, NCNP
- ◆ 1998-2000 Neuromuscular Genetics Post-doc, Department of Neurology, Columbia University

RESEARCH INTERESTS :

- ◆ Muscular dystrophy
- ◆ Congenital myopathy
- ◆ Distal myopathy
- ◆ Metabolic myopathy
- ◆ Muscle pathology

PUBLISHED WORKS : (Selected from 214 PubMed-listed publications)

- Hayashi YK, Matsuda C, Ogawa M, Goto K, Tominaga K, Mitsuhashi S, Park YE, Nonaka I, Hino-Fukuyo N, Haginoya K, Sugano H, Nishino I.
Human PTRF mutations cause secondary deficiency of caveolins resulting in muscular dystrophy with generalized lipodystrophy.
J Clin Invest. 2009 Sep; 119(9):2623-33. Epub 2009 Aug 10.
- Malicdan MC, Noguchi S, Hayashi YK, Nonaka I, Nishino I.
Prophylactic treatment with sialic acid metabolites precludes the development of the myopathic phenotype in the DMRV-hIBM mouse model.
Nat Med. 2009 Jun; 15(6):690-5.
- Ramachandran N, Munteanu I, Wang P, Aubourg P, Rilstone JJ, Israeli N, Naranian T, Paroutis P, Guo R, Ren ZP, Nishino I, Chabrol B, Pellissier JF, Minetti C, Udd B, Fardeau M, Tailor CS, Mahuran DJ, Kissel JT, Kalimo H, Levy N, Manolson MF, Ackerley CA, Minassian BA.
VMA21 deficiency causes an autophagic myopathy by compromising V-ATPase activity and lysosomal acidification.
Cell. 2009 Apr 17; 137(2):235-46.



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- Eisenberg I, Eran A, Nishino I, Moggio M, Lamperti C, Amato AA, Lidov HG, Kang PB, North KN, Mitrani-Rosenbaum S, Flanigan KM, Meely LA, Whitney D, Beggs AH, Kohane IS, Kunkel LM.
Distinctive patterns of microRNA expression in primary muscular disorders.
Proc Natl Acad Sci U S A. 2007 Oct 23; 104(43):17016-21. Epub 2007 Oct 17..
- Malicdan MC, Noguchi S, Nonaka I, Hayashi YK, Nishino I.
A *Gne* knockout mouse expressing human D176V mutation develops features similar to distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy.
Hum Mol Genet. 2007 Nov 15; 16(22):2669-82. Epub 2007 Aug 18.
- Fukami M, Wada Y, Okada M, Nishino I, Hasegawa T, Camerino G, Kretz C, Buj-Bello A, Laporte J, Yamada G, Morohashi K, Ogata T.
CXorf6 is a causative gene for hypospadias.
Nat Genet. 2006 Dec; 38(12):1369-71. Epub 2006 Nov 5.
- Nakagawa O, Arnold M, Nakagawa M, Hamada H, Shelton JM, Kusano H, Harris TM, Childs G, Campbell KP, Richardson JA, Nishino I, Olson EN.
Centronuclear myopathy in mice lacking a novel muscle-specific protein kinase transcriptionally regulated by MEF2.
Genes Dev. 2005 Sep 1; 19(17):2066-77.
- Barresi R, Michele DE, Kanagawa M, Harper HA, Dovico SA, Satz JS, Moore SA, Zhang W, Schachter H, Dumanski JP, Cohn RD, Nishino I, Campbell KP.
LARGE can functionally bypass alpha-dystroglycan glycosylation defects in distinct congenital muscular dystrophies.
Nat Med. 2004 Jul; 10(7):696-703. Epub 2004 Jun 6.
- Noguchi S, Tsukahara T, Fujita M, Kurokawa R, Tachikawa M, Toda T, Tsujimoto A, Arahata K, Nishino I.
cDNA microarray analysis of individual Duchenne muscular dystrophy patients.
Hum Mol Genet. 2003 Mar 15; 12(6):595-600.
- Michele DE, Barresi R, Kanagawa M, Saito F, Cohn RD, Satz JS, Dollar J, Nishino I, Kellely RI, Somer H, Straub V, Mathews KD, Moore SA, Campbell KP.
Posttranslational disruption of dystroglycan-ligand interactions in congenital muscular dystrophies.
Nature. 2002 Jul 25; 418(6896):417-22.
- Nishino I, Fu J, Tanji K, Yamada T, Shimojo S, Koori T, Mora M, Riggs JE, Oh SJ, Koga Y, Sue CM, Yamamoto A, Murakami N, Shanske S, Byrne E, Bonilla E, Nonaka I, DiMauro S, Hirano M.
Primary LAMP-2 deficiency causes X-linked vacuolar cardiomyopathy and myopathy (Danon disease).
Nature. 2000 Aug 24; 406(6798):906-10.
- Papadopoulou LC, Sue CM, Davidson M, Tanji K, Nishino I, Sadlock J, Selby J, Glerum DM, Van Coster R, Lyon G, Scalais E, Lebel R, Kaplan P, Shanske S, De Vivo DC, Bonilla E, Hirano M, DiMauro S, Schon EA.
Fatal infantile cardioencephalomyopathy with COX deficiency and mutations in *SCO2*, a human COX assembly gene.
Nat Genet. 1999 Nov; 23(3):333-7.
- Nishino I, Spinazzola A, Hirano M.
Thymidine phosphorylase gene mutations associated with MNGIE, a human mitochondrial disorder.
Science. 1999 Jan 29; 283(5402):689-92.