A List of Publications

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Date: 17th April 2016

A. Peer-reviewed scientific articles

Original articles in scientific journals

1. **Kilponen JM**, Palosaari PM, Hiltunen JK. Occurrence of a long-chain $\Delta^3\Delta^2$-enoyl-CoA isomerase in rat liver. Biochem J 26(1): 223-226, 1990. (IF 5.016)

2. Palosaari, PM, **Kilponen JM**, Sormunen RT, Hassinen IE, Hiltunen JK. $\Delta^3\Delta^2$-Enoyl-CoA isomerases. Characterization of the mitochondrial isoenzyme in rat. J Biol Chem 265(6): 3347-3353, 1990 (IF 5.328)

3. **Kilponen JM**, Hiltunen JK. β-Oxidation of unsaturated fatty acids in human. Isoenzymes of $\Delta^3\Delta^2$-enoyl-CoA isomerase. FEBS Lett 322: 299-303, 1993. (IF 3.601)

4. **Kilponen JM**, Häyrinen HM, Rehn M, Hiltunen JK. cDNA cloning and amino acid sequence of human mitochondrial $\Delta^3\Delta^2$-enoyl-CoA isomerase. The human enzyme shows dissimilarities with its rat counterpart, mitochondrial short-chain isomerase. Biochem J 300: 1-5, 1994. (IF 5.016)

5. **Uusimaa J**, Vainionpää L, Similä S, Miettinen R, Nuutinen M. L-3-Hydroxyacyl-CoA dehydrogenase deficiency: Two cases with pigmentary retinopathy. J. Inherit. Metab. Dis. 20: 848-850, 1997. (IF 3.6)

6. **Uusimaa J**., Remes AM, Rantala H, Vainionpää L, Herva R, Vuopala K, Nuutinen M, Majamaa K, Hassinen IE. Childhood encephalopathies and myopathies: a prospective study in a defined population to assess the frequency of mitochondrial disorders. Pediatrics. 105(3 Pt 1):598-603, 2000 (IF 5.391)

7. **Uusimaa J**, Finnilä S, Vainionpää L, Kärppä M, Herva R, Rantala H, Hassinen IE, Majamaa K. A Mutation in Mitochondrial DNA-Encoded Cytochrome c Oxidase II Gene in a Child With Alpers- Huttenlocher-like Disease. Pediatrics 111: e262-8, 2003. (IF 5.391)

8. Löppönen T, Väisänen M-L, Luotonen M, Allinen M, **Uusimaa J**, Lindholm P, Mäki-Torkko E, Väyrynen M, Löppönen H, Leisti K. Connexin-26 mutations and non-syndromic hearing impairment: M34T mutation frequency in Northern Finland. Laryngoscope 113: 1758-1763, 2003. (IF 2.096)

9. Ugalde C, Triepels RH, Coenen MJH, van den Heuvel LP, Smeets R, **Uusimaa J**, Briones P, Majamaa K, Smeitink JAM, Nijtmans LGJ. Impaired complex I assembly in a Leigh Syndrome patient with a novel missense mutation in the mitochondrial ND6 gene. Ann Neurol 54(5): 665-669, 2003. (IF 10.746)

10. **Uusimaa J**, Finnilä S, Remes A, Rantala H, Vainionpää L, Hassinen IE, Majamaa K. Molecular Epidemiology of childhood mitochondrial encephalomyopathies in a Finnish populations: sequence analysis of entire mtDNA of 17 children reveals novel heteroplasmic mutations in $tRNA^{Arg}$, $tRNA^{Glu}$ and $tRNA^{Leu(UUR)}$ genes). Pediatrics 114: 443-450,2004. (IF 5.391)

11. Hinttala R, **Uusimaa J**, Remes AM, Rantala H, Hassinen IE, Majamaa K. Sequence analysis of nuclear genes encoding functionally important complex I subunits in children with encephalomyopathy. J Mol Med (Berl). 83:786-94, 2005. (IF 4.77).

12. Laakkonen H, Lönnqvist T, **Uusimaa J**, Qvist E, Valanne L, Nuutinen M, Ala-Houhala M, Majamaa K, Jalanko H, Holmberg C. Muscular dystonia and athetosis in six patients with congenital nephrotic syndrome of the Finnish type (NPHS1).Pediatr Nephrol. 21:182-9, 2006. (IF 2.88)

13. Hinttala R, Smeets R, Moilanen JS, Ugalde C, **Uusimaa J**, Smeitink JA, Majamaa K. Analysis of mitochondrial DNA sequences in patients with isolated or combined oxidative phosphorylation system deficiency. J Med Genet. 43:881-6, 2006. (IF 5.087)

14. Kervinen M, Hinttala R, Helander HM, Kurki S, **Uusimaa J**, Finel M, Majamaa K, Hassinen IE. The MELAS mutations 3946 and 3949 perturb the critical structure in a conserved loop of the ND1 subunit of mitochondrial complex I.Hum Mol Genet.15:2543-52, 2006. (IF 6.67)

15. Ugalde C, Hinttala R, Timal S, Smeets R, Rodenburg RJ, **Uusimaa J**, van Heuvel LP, Nijtmans LG, Majamaa K, Smeitink JA. Mutated ND2 impairs mitochondrial complex I assembly and leads to Leigh syndrome. Mol Genet Metab. 90:10-4, 2007. (IF 2.83)

16. Hinttala R, Karttunen V, Karttunen A, Herva R, **Uusimaa J**, Remes AM. Alexander disease with occipital predominance and a novel c.799G>C mutation in the GFAP gene.Acta Neuropathol. 114:543-5, 2007. (IF 9.78)

17. **Uusimaa J**, Moilanen JS, Vainionpää L, Tapanainen P, Lindholm P, Nuutinen M, Löppönen T, Mäki-Torkko E, Rantala H, Majamaa K. Prevalence, segregation, and phenotype of the mitochondrial DNA 3243A>G mutation in children. Ann Neurol. 62:278-87, 2007. (IF 11.91)

18. **Uusimaa J**, Hinttala R, Rantala H, Päivärinta M, Herva R, Röyttä M, Soini H, Moilanen JS, Remes AM, Hassinen IE, Majamaa K. Homozygous W748S mutation in the POLG1 gene in patients with juvenile-onset Alpers syndrome and status epilepticus.Epilepsia 49:1038-45, 2008. (IF 3.96)

19. Remes AM, Hinttala R, Kärppä M, Soini H, Takalo R, **Uusimaa J**, Majamaa K. Parkinsonism associated with the homozygous W748S mutation in the POLG1 gene.Parkinsonism Relat Disord. 14:652-4, 2008. (IF 4.13)

20. Bolszak M, Anttonen AK, Komulainen T, Hinttala R, Pakanen S, Sormunen R, Herva R, Lehesjoki AE, Majamaa K, Rantala H, **Uusimaa J**. Digenic mutations in severe myoclonic epilepsy of infancy. Epilepsy Res. 85:300-4, 2009. (IF 2.48)

21. Strengell T, Uhari M, Tarkka R, **Uusimaa J**, Alen R, Lautala P, Rantala H. Antipyretic agents for preventing recurrences of febrile seizures: randomized controlled trial.Arch Pediatr Adolesc Med. 2009;163:799-804, 2009. (IF 4.28)

22. Pihlajaniemi TL, Pirttiniemi P, **Uusimaa J**, Majamaa K. Craniofacial morphology in children of mothers with the m.3243A>G mutation in mitochondrial DNA. Cleft Palate Craniofac J. 47: 234-40, 2010.(IF 1.24)

23. O'Toole JF, Liu Y, Davis EE, Westlake CJ, Attanasio M, Otto EA, Seelow D, Nurnberg G, Becker C, Nuutinen M, Kärppä M, Ignatius J, **Uusimaa J**, Pakanen S, Jaakkola E, van den Heuvel LP, Fehrenbach H, Wiggins R, Goyal M, Zhou W, Wolf MT, Wise E, Helou J, Allen SJ, Murga-Zamalloa CA, Ashraf S, Chaki M, Heeringa S, Chernin G, Hoskins BE, Chaib H, Gleeson J, Kusakabe T, Suzuki T, Isaac RE, Quarmby LM, Tennant B, Fujioka H, Tuominen H, Hassinen I, Lohi H, van Houten JL, Rotig A, Sayer JA, Rolinski B, Freisinger P, Madhavan SM, Herzer M, Madignier F, Prokisch H, Nurnberg P, Jackson PK, Khanna H, Katsanis N, Hildebrandt F. Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. J Clin Invest. 120 :791-802, 2010. (IF 12.81)

24. Hinttala R, Kervinen M, **Uusimaa J**, Maliniemi P, Finnilä S, Rantala H, Remes AM, Hassinen IE, Majamaa K. Analysis of functional consequences of haplogroup J polymorphisms m.4216T>C and m.3866T>C in human MT-ND1: mutagenesis of homologous positions in Escherichia coli. Mitochondrion. 10: 358-61, 2010. (IF 3.52)

25. Komulainen T, Hinttala R, Kärppä M, Pajunen L, Finnilä S, Tuominen H, Rantala H, Hassinen I, Majamaa K, **Uusimaa J**. POLG1 p.R722H mutation associated with multiple mtDNA deletions and a neurological phenotype. BMC Neurol. 10: 29, 2010. (IF 2.49)

26. Isohanni P, Hakonen AH, Euro L, Paetau I, Linnankivi T, Liukkonen E, Wallden T, Luostarinen L, Valanne L, Paetau A, **Uusimaa J**, Lönnqvist T, Suomalainen A, Pihko H. POLG1 manifestations in childhood. Neurology 76: 811-5, 2011. (IF 8.30)

27. Suomalainen A, Elo JM, Pietiläinen KH, Hakonen AH, Sevastianova K, Korpela M, Isohanni P, Marjavaara SK, Tyni T, Kiuru-Enari S, Pihko H, Darin N, Õunap K, Kluijtmans LA, Paetau A, Buzkova J, Bindoff LA, Annunen-Rasila J, **Uusimaa J**, Rissanen A, Yki-Järvinen H, Hirano M, Tulinius M, Smeitink J, Tyynismaa H. FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. Lancet Neurol. 10: 806-18, 2011.(IF 21.82)

28. **Uusimaa J**, Jungbluth H, Fratter C, Crisponi G, Feng L, Zeviani M, Hughes I, Treacy EP, Birks J, Brown GK, Sewry CA, McDermott M, Muntoni F, Poulton J. Reversible infantile respiratory chain deficiency is a unique, genetically heterogenous mitochondrial disease. J Med Genet. 48: 660-8, 2011. (IF 5.64)

29. Pätsi J, Maliniemi P, Pakanen S, Hinttala R**, Uusimaa J,** Majamaa K, Nyström T, Kervinen M, Hassinen IE. LHON/MELAS overlap mutation in ND1 subunit of mitochondrial complex I affects ubiquinone binding as revealed by modeling in Escherichia coli NDH-1. Biochim Biophys Acta. 1817: 312-318, 2012.(IF 4.62)

30. Shunmugam S, Hinttala R, Lehtimäki N, Miettinen M, **Uusimaa J**, Majamaa K, Sivonen K, Aro E-M, Mulo P. Nodularia spumigena extract induces upregulation of mitochondrial respiratory chain complexes in spinach (Spinacia oleracea L.) Acta Physiologiae Plantarum 2012.(IF 1.52)

31. Elo JM, Yadavalli SS, Euro L, Isohanni P, Götz A, Carroll CJ, Valanne L, Alkuraya FS, **Uusimaa J**, Paetau A, Caruso EM, Pihko H, Ibba M, Tyynismaa H, Suomalainen A. Mitochondrial phenylalanyl-tRNA synthetase mutations underlie fatal infantile Alpers encephalopathy. Hum Mol Genet. 21:4521-9, 2012. (IF 6.67)

32. Korkiamäki P, Kervinen M, Karjalainen K, Majamaa K, **Uusimaa J**, Remes AM. Prevalence of the primary LHON mutations in Northern Finland associated with bilateral optic atrophy and tobacco-alcohol amblyopia. Acta Ophthalmol. 91: 630-4, 2013. (IF 2.51)

33. **Uusimaa J**, Gowda V, McShane A, Smith C, Evans J, Shrier A, Narasimhan M, O'Rourke A, Rajabally Y, Cowan F, Fratter C, Poulton J. Prospective study of POLG1 mutations presenting in children with intractable epilepsy-prevalence and clinical features. Epilepsia. 54:1002-11, 2013. (IF 4.58).

34. Kervinen M, Widgren P, Saarela V, **Uusimaa J,** Remes A. Leber hereditary optic neuropathy mutations and toxic-genetic optic neuropathy - authors' response. Acta Ophthalmol. 92(1):e78-9, 2014. (IF 2.51)

35. **Uusimaa J**, Evans J, Smith C, Butterworth A, Craig K, Ashley N, Liao C, Kelly D, Samyn M, Rahman S, Stewart H, Morris AAM, Seller A, Fratter C, Taylor RW, Poulton J. Clinical, biochemical, cellular and molecular characterisation of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. Eur J Hum Genet. 22: 184-91, 2014. (IF 4.23)

36. Buler M, Aatsinki SM, Izzi V, **Uusimaa J**, Hakkola J. SIRT5 is under the control of PGC-1α and AMPK and is involved in regulation of mitochondrial energy metabolism. FASEB J. 28: 3225-37, 2014. (IF 5.48)

37. Sofou K, De Coo IF, Isohanni P, Ostergaard E, Naess K, De Meirleir L, Tzoulis C, **Uusimaa J**, De Angst IB, Lönnqvist T, Pihko H, Mankinen K, Bindoff LA, Tulinius M, Darin N. A multicenter study on Leigh syndrome: disease course and predictors of survival. Orphanet J Rare Dis. 9:52, 2014. (IF 3.96)

38. Hynynen J, Komulainen T, Tukiainen E, Nordin A, Arola J, Kälviäinen R, Jutila L, Röyttä M, Hinttala R, Majamaa K, Mäkisalo H, **Uusimaa J**. Acute liver failure after valproate exposure in patients with POLG1 mutations and the prognosis after liver transplantation. Liver Transpl. 20: 1402-12, 2014. (IF 3.79)

39. Komulainen T, Lodge T, Hinttala R, Bolszak M, Pietilä M, Koivunen P, Hakkola J, Poulton J, Morten K, **Uusimaa J**. Sodium valproate induces mitochondrial respiration dysfunction in HepG2 in vitro liver model. Toxicology. 4;331:47-56, 2015. (IF 3.62)

40. Komulainen T, Hautakangas MR, Hinttala R, Pakanen S, Vähäsarja V, Lehenkari P, Olsen P, Vieira P, Saarenpää-Heikkilä O, Palmio J, Tuominen H, Kinnunen P, Majamaa K, Rantala H,

and **Uusimaa J**. Mitochondrial DNA depletion and deletions in paediatric patients with neuromuscular diseases - novel phenotypes. JIMD Rep; 23:91-100, 2015. (IF 4.14)

41. Widgren P, Hurme-Niiranen A, Falck A, Keski-Filppula R, Remes AM, Moilanen J, Majamaa K, Kervinen M, **Uusimaa J**. Genetic etiology of ophthalmological manifestations in children - focus on mitochondrial disease related symptoms. Acta Ophthalmologica Scandinavica, 94:83-91, 2016. (IF 2.844)

42. Carrozzo R, Verrigni D, Rasmussen M, de Coo R, Amartino H, Bianchi M, Buhas D, Mesli S, Naess K, Born AP, Woldseth B, Prontera P, Batbayli M, Ravn K, Joensen F, Cordelli DM, Santorelli FM, Tulinius M, Darin N, Duno M, Jouvencel P, Burlina A, Donti E, Bertin E, Redonnet-Vernhet I, Wibrand F, Dionisi-Vici C, **Uusimaa J**, Vieira P, Osorio A, McFarland R, Taylor RW, Holme E, Ostergaard E. Succinate-CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 25 patients. Accepted for publication, J Inherited Metab Dis, 39:243-52, 2016. (IF 4.14)

43. Diggle CP, Sukoff Rizzo SJ, Popiolek M, Hinttala R, Schülke JP, Kurian MA,Carr IM, Markham AF, Bonthron DT, Watson C, Pysden K, Reinhart V, James LC, Vanase-Frawley MA, Charych E, Allen M, Harms J, Schmidt CJ, Ng J, Strick C, Vieira P, Mankinen K, Kokkonen HL, Kallioinen M, Sormunen R, Rinne JO, Johansson J, Alakurtti K, Huilaja L, Hurskainen T,Tasanen K, Anttila E, Reis Marques T, Howes O, Politis M, Fahiminiya S, Nguyen KQ, Majewski J, **Uusimaa J***, Sheridan E*, Brandon NJ*. Biallelic Mutations in *PDE10A* lead to a loss of striatal PDE10A and a hyperkinetic movement disorder with onset in infancy. Am J Hum Genet 98:735-43, 2016. (IF 10.931). *The shared last authorship.

Reviews in scientific journals

44. Hakkola EH, **Kilponen JM**, Palosaari PM, Hiltunen JK. Pedoksisomitaudit-pediatrista ja neurologista erotusdiagnostiikkaa. Duodecim 107(2): 80-86, 1991

45. Palosaari PM, **Kilponen JM**, Hiltunen JK. Peroxisomal diseases. Ann. Med. 24(3): 163-166, 1992.

46. **Uusimaa J**, Gaily E, Ignatius J, Lehesjoki AE. Current prospects of genetics in epilepsy diagnostics--when and what? Duodecim 125(22):2521-30, 2009.

47. Hakonen AH, Isohanni P, Rantamäki M, Kälviäinen R, Nordin A, **Uusimaa J**, Paetau A, Udd B, Pihko H, Wartiovaara A. Mitochondrial recessive ataxia syndrome (MIRAS) and valproate toxicity. Duodecim. 126(13):1552-9, 2010.

B. Non-refereed scientific articles

48. **Kilponen JM**, Palosaari PM, Sormunen RT, Vihinen M, Hiltunen JK. Isoenzymes of delta 3,delta 2-enoyl-CoA isomerase in rat liver. Prog Clin Biol Res. 375:33-40, 1992

C. Scientific books (monographies)

49. **Kilponen JM**. Metabolism of Δ^3-double bonds of unsaturated fatty acids in mammals. Isoforms of $\Delta^3\Delta^2$-enoyl-CoA isomerase in rat and human. ***Doctoral dissertation, Acta Universitas Ouluensis**, D293, 1994.* ***Original articles 11-14.***

D Publications intended for professional communities

50. **Uusimaa J**, Majamaa K. Mitochondrial diseases. In: Sillanpää, Herrgård, Iivanainen, Koivikko, Rantala (eds) Pediatric neurology, Duodecim, Helsinki, 329-338, 2004.

51. Nuutinen M, **Uusimaa J**. Metabolic diseases. In: Rajantie J, Mertsola J, Heikinheimo M, toim. Lastentaudit. Helsinki: Kustannus Oy Duodecim 2010. (eds). Pediatrics, Duodecim, Helsinki, 2015, in press.

E. Publications intended for the general public, linked to the applicant's research

None.

F Public artistic and design activities

None.

G Theses

52. **Kilponen JM**. Metabolism of Δ^3-double bonds of unsaturated fatty acids in mammals. Isoforms of $\Delta^3\Delta^2$-enoyl-CoA isomerase in rat and human. ***Doctoral dissertation, Acta Universitas Ouluensis**, D293, 1994.* ***Original articles 11-14.***

H Patents and invention disclosures

None.

I Audiovisual material, ICT software

None.