# Tyks kliinisen genetiikan yksikön tieteellisiä julkaisuja - Academic Literature Published by Tyks Department of Clinical Genetics

The wellbeing services county of Southwest Finland

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# 2022

**Detailed prenatal and postnatal MRI findings and clinical analysis of RAF1 in Noonan syndrome.**

Helenius K, Parkkola R, Arola A, Peltola V, **Haanpää MK.**Eur J Med Genet. 2022 Nov;65(11):104626. doi: 10.1016/j.ejmg.2022.104626. Epub 2022 Sep 22.PMID: 36155125

**ATM c.7570G>C is a high-risk allele for breast cancer.**

**Kankuri-Tammilehto M**, Tervasmäki A, Kraatari-Tiri M, Rahikkala E, Pylkäs K, Kuismin O. Int J Cancer. 2022 Sep 26. doi: 10.1002/ijc.34305. Online ahead of print. PMID: 36161273

**Novel germline variant in the histone demethylase and transcription regulator KDM4C induces a multi-cancer phenotype.**

Katainen R, Donner I, Räisänen M, Berta D, Kuosmanen A, Kaasinen E, **Hietala M**, Aaltonen LA. J Med Genet. 2022 Jul;59(7):644-651. doi: 10.1136/jmedgenet-2021-107747. Epub 2021 Jul 19. PMID: 34281993

**Genetic, clinic and histopathologic characterization of BRCA-associated hereditary breast and ovarian cancer in southwestern Finland.**

Pallonen TA, Lempiäinen SMM, Joutsiniemi TK, Aaltonen RI, Pohjola PE, **Kankuri-Tammilehto MK.** Sci Rep. 2022 Apr 25;12(1):6704. doi: 10.1038/s41598-022-10519-y. PMID: 35469032

**Natural history of KBG syndrome in a large European cohort.**

Loberti L, Bruno LP, Granata S, Doddato G, Resciniti S, Fava F, Carullo M, Rahikkala E, Jouret G, Menke LA, Lederer D, Vrielynck P, Ryba L, Brunetti-Pierri N, Lasa-Aranzasti A, Cueto-González AM, Trujillano L, Valenzuela I, Tizzano EF, Spinelli AM, Bruno I, Currò A, Stanzial F, Benedicenti F, Lopergolo D, Santorelli FM, Aristidou C, Tanteles GA, Maystadt I, Tkemaladze T, Reimand T, Lokke H, Õunap K, **Haanpää MK**, Holubová A, Zoubková V, Schwarz M, Žordania R, Muru K, Roht L, Tihveräinen A, Teek R, Thomson U, Isis A, Superti-Furga A, Buoni S, Canitano R, Scandurra V, Rossetti A, Grosso S, Battini R, Baldassarri M, Mencarelli MA, Rizzo CL, Bruttini M, Mari F, Ariani F, Renieri A, Pinto AM. Hum Mol Genet. 2022 Jul 21:ddac167. doi: 10.1093/hmg/ddac167. Online ahead of print. PMID: 35861666

**Clinical and Genetic Characteristics of Finnish Patients with Autosomal Recessive and Dominant Non-Syndromic Hearing Loss Due to Pathogenic TMC1 Variants.**

Kraatari-Tiri M, **Haanpää MK**, Willberg T, Pohjola P, Keski-Filppula R, Kuismin O, Moilanen JS, Häkli S, Rahikkala E. J Clin Med. 2022 Mar 26;11(7):1837. doi: 10.3390/jcm11071837. PMID: 35407445

**Loss-of-function variants in exon 4 of TAB2 cause a recognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletal involvement.**

Micale L, Morlino S, Carbone A, Carissimo A, Nardella G, Fusco C, Palumbo O, Schirizzi A, Russo F, Mazzoccoli G, Breckpot J, De Luca C, Ferraris A, Giunta C, Grammatico P, **Haanpää MK**, Mancano G, Forzano G, Cacchiarelli D, Van Esch H, Callewaert B, Rohrbach M, Castori M. Genet Med. 2022 Feb;24(2):439-453. doi: 10.1016/j.gim.2021.10.009. Epub 2021 Nov 30. PMID: 34906501

**Cytosolic phosphoenolpyruvate carboxykinase deficiency: Expanding the clinical phenotype and novel laboratory findings.**

Vieira P, Nagy II, Rahikkala E, Väisänen ML, Latva K, Kaunisto K, Valmari P, Keski-Filppula R, **Haanpää MK**, Sidoroff V, Miettinen PJ, Arkkola T, Ojaniemi M, Nuutinen M, Uusimaa J, Myllynen P. J Inherit Metab Dis. 2022 Mar;45(2):223-234. doi: 10.1002/jimd.12446. Epub 2021 Nov 11. PMID: 34622459

# 2021

**Thinking outside "The Box": Case-based didactics for medical education and the instructional legacy of Dr John M. Graham, Jr.**

Sanchez-Lara PA, Grand K, **Haanpää MK**, Curry CJ, Wang R, Ezgü F, Rose CM, D'Cunha Burkardt D, Conway RL, Relan A, Carey JC. Am J Med Genet A. 2021 Sep;185(9):2636-2645. doi: 10.1002/ajmg.a.62202. Epub 2021 Apr 29. PMID: 33913595

**Wilsonin tauti - uutta ja vanhaa**

Jussi O.T. Sipilä, **Marja Hietala** ja Valtteri Kaasinen
Duodecim 2021;137(17):1767-73
Katsaus. In Finnish.
Lehti 17/2021

**Perinnöllisyyslääkärin osuus syövän geenidiagnostiikassa - kokemukset Tyksistä ja muualta**

**Minna Kankuri-Tammilehto**, Minna Pöyhönen ja **Maria Haanpää**
Duodecim 2021;137(13):1449-56
Teema: Geeniohjatun syövän hoidon työryhmä. In Finnish.
Lehti 13/2021

**An ARHGAP25 variant links aberrant Rac1 function to early-onset skeletal fragility.**

Mäkitie RE, Henning P, Jiu Y, Kämpe A, Kogan K, Costantini A, Välimäki VV, Medina-Gomez C, Pekkinen M, Salusky IB, Schalin-Jäntti C, **Haanpää MK**, Rivadeneira F, Bassett JHD, Williams GR, Lerner UH, Pereira RC, Lappalainen P, Mäkitie O. JBMR Plus. 2021 Jun 7;5(7):e10509. doi: 10.1002/jbm4.10509. eCollection 2021 Jul. PMID: 34258505

**Geeniohjatun syövän hoidon työryhmä hoitopäätösten apuna**

Erika Alanne, Katri Orte, **Maria Haanpää**, **Minna Kankuri-Tammilehto**, Maria Silvoniemi, Maria Sundvall, Sakari Hietanen, Pia Boström, Jukka Laine, Panu Jaakkola, Heikki Minn, Markku Kallajoki, Veli Kairisto, Klaus Elenius ja Pia Vihinen

Duodecim 2021;137(13):1437-40

Teema: Geeniohjatun syövän hoidon työryhmä. In Finnish.

Lehti 13/2021

**Neurocognitive follow-up in adult siblings with Phelan-McDermid syndrome due to a novel SHANK3 splicing site mutation.**

**Kankuri-Tammilehto M**, Sauna-Aho O, Arvio M. Mol Genet Genomic Med. 2021 Dec;9(12):e1780. doi: 10.1002/mgg3.1780. Epub 2021 Aug 9. PMID: 34369668

**Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland.**

Järvelä I, Määttä T, Acharya A, Leppälä J, Jhangiani SN, Arvio M, Siren A, **Kankuri-Tammilehto M**, Kokkonen H, Palomäki M, Varilo T, Fang M, Hadley TD, Jolly A, Linnankivi T, Paetau R, Saarela A, Kälviäinen R, Olme J, Nouel-Saied LM, Cornejo-Sanchez DM, Llaci L, Lupski JR, Posey JE, Leal SM, Schrauwen I. Hum Genet. 2021 Jul;140(7):1011-1029. doi: 10.1007/s00439-021-02268-1. Epub 2021 Mar 12. PMID: 33710394

**Novel germline variant in the histone demethylase and transcription regulator KDM4C induces a multi-cancer phenotype.**

Katainen R, Donner I, Räisänen M, Berta D, Kuosmanen A, Kaasinen E, **Hietala M**, Aaltonen LA. J Med Genet. 2022 Jul;59(7):644-651. doi: 10.1136/jmedgenet-2021-107747. Epub 2021 Jul 19. PMID: 34281993

## 2020

**Wilson's Disease in Finland: A Nationwide Population-Based Study.**

Sipilä JOT, **Hietala M**, Kytö V, Kaasinen V. Mov Disord. 2020 Dec;35(12):2323-2327. doi: 10.1002/mds.28207. Epub 2020 Jul 2. PMID: 32618023

**Genetic Susceptibility to Kidney Cancer | IntechOpen**

**Minna Kankuri-Tammilehto**

Submitted: May 7th, 2019 Reviewed: February 28th, 2020 Published: June 2nd, 2020

DOI: 10.5772/intechopen.91933

## 2019

**Natural course of Fabry disease with the p. Arg227Ter (p.R227*) mutation in Finland: Fast study.**

Pietilä-Effati P, Saarinen JT, Löyttyniemi E, Autio R, Saarenhovi M, **Haanpää MK**, Kantola I.

Mol Genet Genomic Med. 2019 Oct;7(10):e00930. doi: 10.1002/mgg3.930. Epub 2019 Aug 14.

**Two middle-aged women with the Finnish variant of muscle-eye-brain disease (MEB).**

Arvio M, Määttänen L, **Haanpää M**, Lähdetie J

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**High-Resolution Bisulfite-Sequencing of Peripheral Blood DNA Methylation in Early-Onset and Familial Risk Breast Cancer Patients.**

Chen J, **Haanpää MK**, Gruber JJ, Jäger N, Ford JM, Snyder MP. Clin Cancer Res. 2019 Sep 1;25(17):5301-5314. doi: 10.1158/1078-0432.CCR-18-2423. Epub 2019 Jun 7. PMID: 31175093

**HTT haplogroups in Finnish patients with Huntington disease.**

Ylönen S, Sipilä JOT, **Hietala M**, Majamaa K. Neurol Genet. 2019 Apr 22;5(3):e334. doi: 10.1212/NXG.0000000000000334. eCollection 2019 Jun. PMID: 31086827

**Huntingtonin tauti**

Jussi O. T. Sipilä, Valtteri Kaasinen, **Marja Hietala**, Markku Päivärinta ja Kari Majamaa

Duodecim 2019;135(3):249-5. Katsausartikkeli. In Finnish.

**Syövän perinnöllisyys**

**Kankuri-Tammilehto M,** Vihinen P, Schleutker J

Lääkärilehti 14/2019 s 30-36. Katsausartikkeli. In Finnish.

**Keskustelu on tärkeää tiedon määrän kasvaessa**

Lehti 17: Keskustelua 26.4.2019 17/2019 vsk 74 s. 1027. In Finnish.

**Minna Kankuri-Tammilehto**, Pia Vihinen, Johanna Schleutker

**ALG11-CDG syndrome: Expanding the phenotype.**

**Haanpää MK**, Ng BG, Gallant NM, Singh KE, Brown C, Kimonis V, Freeze HH, Muller EA 2nd. Am J Med Genet A. 2019 Mar;179(3):498-502. doi: 10.1002/ajmg.a.61046. Epub 2019 Jan 24. PMID: 30676690

**Hypomorphic mutations of TRIP11 cause odontochondrodysplasia.**

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JCI Insight. 2019 Feb 7;4(3). pii: 124701. doi: 10.1172/jci.insight.124701. [Epub ahead of print]

# 2018

**Clinical and genetic characteristics of late-onset Huntington's disease.**

Oosterloo M, Bijlsma EK, van Kuijk SM, Minkels F, de Die-Smulders CE; REGISTRY Investigators of the European Huntington's Disease Network (Bachoud-Lévi AC, Bentvoglio AR…Peippo M, Sipponen M, Bruun A, Hartikainen P, Mäkipää S, Ollokainen M, Åman J, Kärppä M, **Ignatius J**…Pica E, Roos RA); Registry Steering committee; Language coordinators; EHDN's associate site in Singapore.

Parkinsonism Relat Disord. 2018 Nov 29. pii: S1353-8020(18)30490-5. doi: 10.1016/j.parkreldis.2018.11.009. [Epub ahead of print]

**Recessive multiple epiphyseal dysplasia - Clinical characteristics caused by rare compound heterozygous SLC26A2 genotypes.**

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Eur J Med Genet. 2018 Nov 10. pii: S1769-7212(18)30181-2. doi: 10.10 16/j.ejmg.2018.11.007. [Epub ahead of print]

**Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes.**

Vos JR, Giepmans L, Röhl C, Geverink N, Hoogerbrugge N; ERN GENTURIS (Hoogerbrugge N, Ligtenberg M…Peltonen S, **Hietala M**, Colas C).

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**Placenta-mediated pregnancy complications are not associated with fetal or paternal factor V Leiden mutation.**

Nevalainen J**, Ignatius J**, Savolainen ER, Ryynanen M, Järvenpaa J.

Eur J Obstet Gynecol Reprod Biol. 2018 Nov;230:32-35. doi: 10.1016/j.ejogrb.2018.09.016. Epub 2018 Sep 11.

**Some DNM2 mutations cause extremely severe congenital myopathy and phenocopy myotubular myopathy.**

Biancalana V, Romero NB, Thuestad IJ, **Ignatius J**, Kataja J, Gardberg M, Héron D, Malfatti E, Oldfors A, Laporte J.

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**Sodium Channel SCN3A (NaV1.3) Regulation of Human Cerebral Cortical Folding and Oral Motor Development.**

Smith RS, Kenny CJ, Ganesh V, Jang A, Borges-Monroy R, Partlow JN, Hill RS, Shin T, Chen AY, Doan RN, Anttonen AK, **Ignatius J**, Medne L, Bönnemann CG, Hecht JL, Salonen O, Barkovich AJ, Poduri A, Wilke M, de Wit MCY, Mancini GMS, Sztriha L, Im K, Amrom D, Andermann E, Paetau R, Lehesjoki AE, Walsh CA, Lehtinen MK.

Neuron. 2018 Sep 5;99(5):905-913.e7. doi: 10.1016/j.neuron.2018.07.052. Epub 2018 Aug 23.

**Suicidal ideation and suicidal behavior according to the C-SSRS in a European cohort of Huntington's disease gene expansion carriers.**

van Duijn E, Vrijmoeth EM, Giltay EJ, Bernhard Landwehrmeyer G; REGISTRY investigators of the European Huntington's Disease Network (Bachoud-Lévi AC, Bentivoglio AR…Peippo M, Sipponen M, **Ignatius J**…Pica E, Roos RA).

J Affect Disord. 2018 Mar 1;228:194-204. doi: 10.1016/j.jad.2017.11.074. Epub 2017 Nov 15.

**Reduced Cancer Incidence in Huntington's Disease: Analysis in the Registry Study.**

McNulty P, Pilcher R, Ramesh R, Necuiniate R, Hughes A, Farewell D, Holmans P, Jones L; REGISTRY Investigators of the European Huntington's Disease Network (Bonelli RM, Hecht K…Peippo M, Sipponen M, Hartikainen P, Ollokainen M, Åman J, **Ignatius J**…Yudina E, Zielonka E).

J Huntingtons Dis. 2018;7(3):209-222. doi: 10.3233/JHD-170263.

**Abnormal coagulation and enhanced fibrinolysis due to lysinuric protein intolerance associates with bleeds and renal impairment.**

Pitkänen HH, Kärki M, Niinikoski H, **Tanner L**, Näntö-Salonen K, Pikta M, Kopatz WF, Zuurveld M, Meijers JCM, Brinkman HJM, Lassila R.

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**A 69-year-old woman with Coffin-Siris syndrome.**

Määttänen L, **Hietala M, Ignatius J**, Arvio M.

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**A homozygous I684T in GLE1 as a novel cause of arthrogryposis and motor neuron loss.**

Paakkola T, Vuopala K, Kokkonen H, **Ignatius J,** Valkama M, Moilanen JS, Fahiminiya S, Majewski J, Hinttala R, Uusimaa J.

Clin Genet. 2018 Jan;93(1):173-177. doi: 10.1111/cge.13086. Epub 2017 Nov 24.

# 2017

**A homozygous FANCM mutation underlies a familial case of non-syndromic primary ovarian insufficiency.**

Fouquet B, Pawlikowska P, Caburet S, Guigon C, Mäkinen M, **Tanner L, Hietala M**, Urbanska K, Bellutti L, Legois B, Bessieres B, Gougeon A, Benachi A, Livera G, Rosselli F, Veitia RA, Misrahi M.

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**Novel COL1A1 Mutation c.3290G>T Associated With Severe Form of Osteogenesis Imperfecta in a Fetus.**

**Tanner L**, Vainio P, Sandell M, Laine J.

Pediatr Dev Pathol. 2017 Sep-Oct;20(5):455-459. doi: 10.1177/1093526616686903. Epub 2017 Jan 25.

**Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues.**

Biancalana V, Scheidecker S, Miguet M, Laquerrière A, Romero NB, Stojkovic T, Abath Neto O, Mercier S, Voermans N, **Tanner L**, Rogers C, Ollagnon-Roman E, Roper H, Boutte C, Ben-Shachar S, Lornage X, Vasli N, Schaefer E, Laforet P, Pouget J, Moerman A, Pasquier L, Marcorelle P, Magot A, Küsters B, Streichenberger N, Tranchant C, Dondaine N, Schneider R, Gasnier C, Calmels N, Kremer V, Nguyen K, Perrier J, Kamsteeg EJ, Carlier P, Carlier RY, Thompson J, Boland A, Deleuze JF, Fardeau M, Zanoteli E, Eymard B, Laporte J.

Acta Neuropathol. 2017 Dec;134(6):889-904. doi: 10.1007/s00401-017-1748-0. Epub 2017 Jul 6.

**Germline EMSY sequence alterations in hereditary breast cancer and ovarian cancer families.**

Määttä KM, Nurminen R, **Kankuri-Tammilehto M**, Kallioniemi A, Laasanen SL, Schleutker J.

BMC Cancer. 2017 Jul 24;17(1):496. doi: 10.1186/s12885-017-3488-x.

**SNCA mutation p.Ala53Glu is derived from a common founder in the Finnish population.**

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Neurobiol Aging. 2017 Feb;50:168.e5-168.e8. doi: 10.1016/j.neurobiolaging.2016.10.014. Epub 2016 Oct 19.

**Inhaled Sargramostim Induces Resolution of Pulmonary Alveolar Proteinosis in Lysinuric Protein Intolerance.**

**Tanner LM**, Kurko J, Tringham M, Aho H, Mykkänen J, Näntö-Salonen K, Niinikoski H, Lukkarinen H.

JIMD Rep. 2017;34:97-104. doi: 10.1007/8904_2016_15. Epub 2016 Oct 26.

**Food allergy in a child with de novo KAT6A mutation.**

Elenius V, Lähdesmäki T, **Hietala M**, Jartti T.

Clin Transl Allergy. 2017 Jun 22;7:19. doi: 10.1186/s13601-017-0155-x. eCollection 2017

**TUFT1, a novel candidate gene for metatarsophalangeal osteoarthritis, plays a role in chondrogenesis on a calcium-related pathway.**

Sliz E, Taipale M, Welling M, Skarp S, Alaraudanjoki V, **Ignatius J**, Ruddock L, Nissi R, Männikkö M.

PLoS One. 2017 Apr 14;12(4):e0175474. doi: 10.1371/journal.pone.0175474. eCollection 2017.

**Interrupted/bipartite clavicle as a diagnostic clue in Kabuki syndrome.**

**Haanpää M**, Schlecht H, Batra G, Clayton-Smith J, Douzgou S.

Am J Med Genet A. 2017 Apr;173(4):1115-1118. doi: 10.1002/ajmg.a.38131. Epub 2017 Mar 3.

**TSC2 c.1864C>T variant associated with mild cases of tuberous sclerosis complex.**

Farach LS, Gibson WT, Sparagana SP, Nellist M, Stumpel CT, **Hietala M**, Friedman E, Pearson DA, Creighton SP, Wagemans A, Segel R, Ben-Shalom E, Au KS, Northrup H.

Am J Med Genet A. 2017 Mar;173(3):771-775. doi: 10.1002/ajmg.a.38083. Review.

**Geneettinen alttius syövälle**

**Minna Kankuri-Tammilehto** ja Johanna Schleutker

Duodecim 2017;133(19):1773-81. Katsausartikkeli. In Finnish.

# 2016

**Imbalance of plasma amino acids, metabolites and lipids in patients with lysinuric protein intolerance (LPI).**

Kurko J, Tringham M, **Tanner L**, Näntö-Salonen K, Vähä-Mäkilä M, Nygren H, Pöhö P, Lietzen N, Mattila I, Olkku A, Hyötyläinen T, Orešič M, Simell O, Niinikoski H, Mykkänen J.

Metabolism. 2016 Sep;65(9):1361-75. doi: 10.1016/j.metabol.2016.05.012. Epub 2016 May 27.

**Skeletal Characteristics of WNT1 Osteoporosis in Children and Young Adults.**

Mäkitie RE, **Haanpää M**, Valta H, Pekkinen M, Laine CM, Lehesjoki AE, Schalin-Jäntti C, Mäkitie O.

J Bone Miner Res. 2016 Sep;31(9):1734-42. doi: 10.1002/jbmr.2841. Epub 2016 Apr 25.

**Whole-exome sequencing of Finnish hereditary breast cancer families.**

Määttä K, Rantapero T, Lindström A, Nykter M, **Kankuri-Tammilehto M**, Laasanen SL, Schleutker J.

Eur J Hum Genet. 2017 Jan;25(1):85-93. doi: 10.1038/ejhg.2016.141. Epub 2016 Oct 26.

**Detection of Novel Gene Variants Associated with Congenital Hypothyroidism in a Finnish Patient Cohort.**

Löf C, Patyra K, Kuulasmaa T, Vangipurapu J, Undeutsch H, Jaeschke H, Pajunen T, Kero A, Krude H, Biebermann H, Kleinau G, Kühnen P, Rantakari K, Miettinen P, Kirjavainen T, Pursiheimo JP, Mustila T, Jääskeläinen J, Ojaniemi M, Toppari J, **Ignatius J**, Laakso M, Kero J.

Thyroid. 2016 Sep;26(9):1215-24. doi: 10.1089/thy.2016.0016. Epub 2016 Aug 2.

**A novel transthyretin Lys70Glu (p.Lys90Glu) mutation presenting with vitreous amyloidosis and carpal tunnel syndrome.**

Raivio VE, Jonasson J, Myllykangas L, Ala-Mello S, **Kankuri-Tammilehto M**, Kiuru-Enari S, Westermark P, Tanskanen M, Kivelä T.

Amyloid. 2016;23(1):46-50. doi: 10.3109/13506129.2015.1126574. Epub 2016 Feb 1.

**Lääkärin työpäivä vuonna 2036**

**Minna Kankuri-Tammilehto**, Kirsi Launonen, Jarno Tuominen ja Marko Ahvenainen

Duodecim 2016;132(23):2193-7. In Finnish.

**Urine Beta2-Microglobulin Is an Early Marker of Renal Involvement in LPI.**

Kärki M, Näntö-Salonen K, Niinikoski H, **Tanner LM**.

JIMD Rep. 2016;25:47-55. Epub 2015 Jun 30.

**Lastentaudit ja periytyminen.**

**Penttinen M**, **Hietala M**.

Kirjassa: Lastentaudit. Rajantie J, Heikinheimo M, Renko M (toim.). ss. 79-90. Kustannus Oy Duodecim, Helsinki 2016. 6. uudistettu painos. In Finnish.

**Geneettiset seulonnat**

**Hietala M**, Lapatto R.

Kirjassa: Lääketieteellinen genetiikka. K. Aittomäki, J. Moilanen, M. Perola (toim.) Kustannus Oy Duodecim, Helsinki 2016. 1. painos. In Finnish.

# 2015

**Dysfunction in macrophage toll-like receptor signaling caused by an inborn error of cationic amino acid transport.**

Kurko J, Vähä-Mäkilä M, Tringham M, **Tanner L**, Paavanen-Huhtala S, Saarinen M, Näntö-Salonen K, Simell O, Niinikoski H, Mykkänen J.

Mol Immunol. 2015 Oct;67(2 Pt B):416-25. doi: 10.1016/j.molimm.2015.07.006. Epub 2015 Jul 22.

**Clinical and imaging findings in Parkinson disease associated with the A53E SNCA mutation.**

Martikainen MH, Päivärinta M, **Hietala M**, Kaasinen V.

Neurol Genet. 2015 Oct 15;1(4):e27. doi: 10.1212/NXG.0000000000000027. eCollection 2015 Dec.

**PARK2-geenin mutaatioon liittyvä Parkinsonin tauti**

Valtteri Kaasinen, **Marja Hietala** ja Mikko Kuoppamäki

Duodecim 2015;131(12):1187-90

Tapausselostus. In Finnish.

**Epidemiology of Huntington's disease in Finland.**

Sipilä JO, **Hietala M**, Siitonen A, Päivärinta M, Majamaa K.

Parkinsonism Relat Disord. 2015 Jan;21(1):46-9. doi: 10.1016/j.parkreldis.2014.10.025. Epub 2014 Nov 3.

# 2014

**Molecular analysis of two novel missense mutations in the GDF5 proregion that reduce protein activity and are associated with brachydactyly type C.**

Stange K, Thieme T, Hertel K, Kuhfahl S, Janecke AR, Piza-Katzer H, **Penttinen M**, **Hietala M**, Dathe K, Mundlos S, Schwarz E, Seemann P.

J Mol Biol. 2014 Sep 23;426(19):3221-3231. doi: 10.1016/j.jmb.2014.07.029. Epub 2014 Aug 1.

# 2013

**Suicidal ideation in a European Huntington's disease population.**

Hubers AA, van Duijn E, Roos RA, Craufurd D, Rickards H, Bernhard Landwehrmeyer G, van der Mast RC, Giltay EJ; REGISTRY investigators of the European Huntington's Disease Network (Bachoud-Lévi AC, Bentivoglio AR…Peippo M, **Ignatius J**…).
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