CURRICULUM VITAE, Giedre Grigelioniene

EMPLOYMENT

April 2015 – July 2016	Visiting Scientist, Endocrine lab, Massachusetts General Hospital, Boston, USA.
September 2012–present	Consultant in Clinical Genetics and Pediatrics, Center for Rare Diseases, Department of Clinical Genetics, Karolinska University Hospital Solna, Stockholm, Sweden.
July 2005–September 2012	Resident physician in Pediatrics and Clinical Genetics, Karolinska University Hospital Solna, Stockholm, Sweden.
November 2001–June 2005	Research internship, Karolinska Hospital, Stockholm, Sweden.
July–December 1994	Internship at Vilnius University Hospital, Lithuania.
January–December 1993	Nurse (part time), Department of Vascular Surgery, Vilnius University Hospital, Lithuania.
January–December 1992	Nurse (part time), Department of Neonatology, Vilnius University Hospital, Lithuania.

EDUCATION

September 6, 2012	Specialist in genetics, expertise in congenital skeletal disorders since 1995.
July 17, 2012	Specialist in pediatrics.
July 21, 2005	Swedish medical licence.
September 28, 2001	PhD in pediatric science, thesis “Clinical and Genetic Investigation of Hypochondroplasia and Dyschondrosteosis”.
February 15, 2001	Medical exam for physicians qualified outside EU/EEA (TULE), Karolinska Institute, Stockholm, Sweden.
November 23, 1999	Swedish language exam for physicians qualified outside EU/EEA.
January 1996–Sept 2001	PhD program, Department of Pediatric Endocrinology, Karolinska Hospital, Stockholm, Sweden.
January–December 1995	Research Scholarship for Young Investigators, European Society for Pediatric Research, Department of Neonatology, Karolinska Hospital, Stockholm, Sweden.
July 1, 1994	Physician *Diploma cum Laude*, Vilnius University, Lithuania.
September 1988–July 1994	Student, Faculty of Medicine, Vilnius University, Lithuania.

MEMBERSHIP & ACTIVITIES IN PROFESSIONAL ASSOCIATIONS

Since 2014 member of American Society of Human Genetics
Since 2013 member of European Society for Pediatric Endocrinology
Since 2012 member of International Skeletal Dysplasia Network
Since 2012 member of the Board of Swedish Pediatric Genetics Association
Since 2000 member of Swedish Society of Medicine

VOLUNTEER WORK

April 2014	Review of the report on achodrogenesis type IA and autosomal recessive multiple epiphyseal dysplasia for National Organization for Rare Disorders (NORD)
2007–2011	Assistant at German Short Stature People Meeting (Kleinwuchsforum), organized by German Short Stature People Federal Association (Bundesverband Kleinwüchsige Mensche und ihre Familien e. V.) in Hohenroda, Germany

PUBLICATIONS

Publications during the last 8 years:
Total number of publications during last 8 years 40.

Peer-reviewed original articles:

1. Suzuki HI, Sprengler RM, **Grigelioniene G**, Kobayashi T, Sharp PA. Deconvolution of seed and RNA-binding protein crosstalking RNAi based functional genomics. Nat Genet, 2018, *In Press.*
2. Leal GF, Nishimura G, Voss U, Bertola DR, Åström E, Svensson J, Yamamoto GL, Hammarsjö A, Horemuzova E, Papadiogannakis N, Iwarsson E, **Grigelioniene G#**, Tham E#. Expanding the clinical spectrum of phenotypes caused by pathogenic variants in *PLOD2*. J Bone Miner Res. 2017 Nov 27. [Epub ahead of print]. PubMed PMID: 29178448. # equal contribution.
3. Hammarsjö A, Wang Z, Vaz R, Taylan F, Sedghi M, Girisha KM, Chitayat D, Neethukrishna K, Shannon P, Godoy R, Gowrishankar K, Lindstrand A, Nasiri J, Baktashian M, Newton PT, Guo L, Hofmeister W, Pettersson M, Chagin AS, Nishimura G, Yan L, Matsumoto N, Nordgren A, Miyake N, **Grigelioniene G#**, Ikegawa S#. Novel *KIAA0753* mutations extend the phenotype of skeletal ciliopathies. Sci Rep. 2017 Nov 14;7(1):15585. PubMed PMID: 29138412. # equal contribution.
4. Pekkinen M, **Grigelioniene G**, Akin L, Shah K, Karaer K, Kurtoğlu S, Ekbote A, Aycan Z, Sağsak E, Danda S, Åström E, Mäkitie O. Novel mutations in the *LRP5* gene in patients with osteoporosis-pseudoglioma syndrome. Am J Med Genet A. 2017 Dec;173(12):3132-3135. PubMed PMID:29055141.
5. Andersson K, Dahllöf G, Lindahl K, Kindmark A, **Grigelioniene G**, Åström E, Malmgren B. Mutations in *COL1A1* and *COL1A2* and dental aberations in children and adolescents with osteogenesis imperfecta-a retrospective cohort study. PLoS One. 2017 May 12;12(5):e0176466. PubMed PMID: 28498836.
6. Wang Z, Horemuzova E, Iida A, Guo L, Liu Y, Matsumoto N, Nishimura G, Nordgren A, Miyake N, Tham E, **Grigelioniene G**, Ikegawa S. Axial spondylometaphyseal dysplasia is also caused by *NEK1* mutations. J Hum Genet. 2017 Apr;62(4):503-506. PubMed PMID:28123176.
7**. Grigelioniene G**, Nevalainen PI, Reyes M, Thiele S, Tafaj O, Molinaro A, Takatani R, Nilsson D, Eisfeldt J, Lindstrand A, Kottler ML, Mäkitie O, Jüppner H. A large inversion involving *GNAS* exon A/B and all exons encoding Gsα is associated with autosomal dominant pseudohypoparathyroidism type Ib (PHP1B). J Bone Miner Res. 2017 Apr;32(4):776-783. PubMed PMID:28084650.
8. Malmgren B, Andersson K, Lindahl K, Kindmark A, **Grigelioniene G**, Zachariadis V, Dahllöf G, Åström E. Tooth agenesis in osteogenesis imperfecta related to mutations in the collagen type I genes. Oral Dis. 2017 Jan; 23(1):42-49 Aug 11. PubMed PMID:27510842.
9. Handa A, Tham E, Wang Z, Horemuzova E, **Grigelioniene G**. Autosomal recessive brachyolmia: early radiological findings. Skeletal Radiol. 2016 Nov;45(11):1557-60. PubMed PMID:27544198.
10. Taylan F, Costantini A, Coles N, Pekkinen M, Héon E, Şıklar Z, Berberoğlu M, Kämpe A, Kıykım E, **Grigelioniene G**, Tüysüz B, Mäkitie O. Spondyloocular Syndrome - Novel Mutations in XYLT2 Gene and Expansion of the Phenotypic Spectrum. J Bone Miner Res. 2016 Mar 14. doi: 10.1002/jbmr.2834. 2016;31(8):1577-85. PubMed PMID: 26987875.
11. Wang Z, Iida A, Miyake N, Nishiguchi KM, Fujita K, Nakazawa T, Alswaid A, Albalwi MA, Kim OH, Cho TJ, Lim GY, Isidor B, David A, Rustad CF, Merckoll E, Westvik J, Stattin EL, **Grigelioniene G**, Kou I, Nakajima M, Ohashi H, Smithson S, Matsumoto N, Nishimura G, Ikegawa S. Axial Spondylometaphyseal Dysplasia Is Caused by *C21orf2* Mutations. PLoS One. 2016 Mar 14;11(3):e0150555. doi: 10.1371/journal.pone.0150555. eCollection 2016. PubMed PMID: 26974433.
12. Lindahl K, Kindmark A, Rubin CJ, Malmgren B, **Grigelioniene G**, Söderhäll S, Ljunggren Ö, Åström E. Decreased fracture rate, pharmacogenetics and BMD response in 79 Swedish children with osteogenesis imperfecta types I, III and IV treated with Pamidronate. Bone. 2016 Mar 5. 2016;87:11-8. doi: 10.1016/j.bone.2016.02.015. PubMed PMID: 26957348.
13. Kuchinskaya E#, **Grigelioniene G#**, Hammarsjö A, Lee HR, Högberg L, Grigelionis G, Kim OH, Nishimura G, Cho TJ. Extending the phenotype of *BMPER*-related skeletal dysplasias to ischiospinal dysostosis. Orphanet J Rare Dis. 2016 Jan 4;11(1):1. doi: 10.1186/s13023-015-0380-0. PubMed PMID: 26728142. # equal contribution.
14. Takatani R, Molinaro A, **Grigelioniene G,** Tafaj O, Watanabe T, Reyes M, Sharma A, Singhal V, Raymond FL, Linglart A, Jüppner H. Analysis of Multiple Families With Single Individuals Affected by Pseudohypoparathyroidism Type Ib (PHP1B) Reveals Only One Novel Maternally Inherited *GNAS* Deletion. J Bone Miner Res. 2015 Oct 19. doi: 10.1002/jbmr.2731. 2016;31(4):796-805. PubMed PMID: 26479409.
15. Hammarsjö A, Nordgren A, Lagerstedt-Robinson K, Malmgren H, Nilsson D, Wedrén S, Nordenskjöld M, Nishimura G, **Grigelioniene G**. Pathogenenic variant in the *COL2A1* gene is associated with Spondyloepiphyseal dysplasia type Stanescu. Am J Med Genet A. 2016 Jan;170(1):266-9. doi: 10.1002/ajmg.a.37387. Epub 2015 Sep 30. PubMed PMID: 26420734.

16. Ockeloen CW, Willemsen MH, de Munnik S, van Bon BW, de Leeuw N, Verrips A, Kant SG, Jones EA, Brunner HG, van Loon RL, Smeets EE, van Haelst MM, van Haaften G, Nordgren A, Malmgren H, **Grigelioniene G**, Vermeer S, Louro P, Ramos L, Maal TJ, van Heumen CC, Yntema HG, Carels CE, Kleefstra T. Further delineation of the KBG syndrome caused by ANKRD11 aberrations. Eur J Hum Genet. 2015 Sep;23(9):1270. doi: 10.1038/ejhg.2015.130. PubMed PMID: 26269249.
17. Lindahl K, Åström E, Rubin CJ, **Grigelioniene G**, Malmgren B, Ljunggren Ö, Kindmark A. Genetic epidemiology, prevalence, and genotype-phenotype correlations in the Swedish population with osteogenesis imperfecta. Eur J Hum Genet. 2015 Aug;23(8):1112. doi: 10.1038/ejhg.2015.129. PubMed PMID: 26177859.
18. Tham E, Eklund EA, Hammarsjö A, Bengtson P, Geiberger S, Lagerstedt-Robinson K, Malmgren H, Nilsson D, Grigelionis G, Conner P, Lindgren P, Lindstrand A, Wedell A, Albåge M, Zielinska K, Nordgren A, Papadogiannakis N, Nishimura G, **Grigelioniene G.** A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbach-Nishimura skeletal dysplasia due to pathogenic variants in *ALG9*. Eur J Hum Genet. 2016 Feb;24(2):198-207. doi: 10.1038/ejhg.2015.91. Epub 2015 May 13. PubMed PMID: 25966638.
19. Reynaert N, Ockeloen CW, Sävendahl L, Beckers D, Devriendt K, Kleefstra T, Carels CE, **Grigelioniene G**, Nordgren A, Francois I, de Zegher F, Casteels K. Short Stature in KBG Syndrome: First Responses to Growth Hormone Treatment. Horm Res Paediatr. 2015;83(5):361-4. doi: 10.1159/000380908. Epub 2015 Apr 1. PubMed PMID: 25833229.
20. Laurell T, Nilsson D, Hofmeister W, Lindstrand A, Ahituv N, Vandermeer J, Amilon A, Annerén G, Arner M, Pettersson M, Jäntti N, Rosberg HE, Cattini PA, Nordenskjöld A, Mäkitie O, **Grigelioniene G,** Nordgren A. Identification of three novel *FGF16* mutations in X-linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. Mol Genet Genomic Med. 2014 Sep;2(5):402-11. doi: 10.1002/mgg3.81. Epub 2014 May 14. PubMed PMID:25333065.
21. Acuna-Hidalgo R, Schanze D, Kariminejad A, Nordgren A, Kariminejad MH, Conner P, **Grigelioniene G**, Nilsson D, Nordenskjöld M, Wedell A, Freyer C, Wredenberg A, Wieczorek D, Gillessen-Kaesbach G, Kayserili H, Elcioglu N, Ghaderi-Sohi S, Goodarzi P, Setayesh H, van de Vorst M, Steehouwer M, Pfundt R, Krabichler B, Curry C, MacKenzie MG, Boycott KM, Gilissen C, Janecke AR, Hoischen A, Zenker M. Neu-Laxova syndrome is a heterogeneous metabolic disorder caused by defects in enzymes of the L-serine biosynthesis pathway. Am J Hum Genet. 2014 Sep 4;95(3):285-93. doi: 10.1016/j.ajhg.2014.07.012. Epub 2014 Aug 21. PubMed PMID: 25152457.
23. Tham E, Nishimura G, Geiberger S, Horemuzova E, Nilsson D, Lindstrand A, Hammarsjö A, Armenio M, Mäkitie O, Zabel B, Nordgren A, Nordenskjöld M, **Grigelioniene G**. Autosomal recessive mutations in the *COL2A1* gene cause severe spondyloepiphyseal dysplasia. Clin Genet. 2015 May;87(5):496-8. doi: 10.1111/cge.12466. Epub 2014 Sep 8. PubMed PMID: 25060605.
24. **Grigelioniene G**, Geiberger S, Horemuzova E, Moström E, Jäntti N, Neumeyer L, Åström E, Nordenskjöld M, Nordgren A, Mäkitie O. Autosomal dominant brachyolmia in a large Swedish family: phenotypic spectrum and natural course. Am J Med Genet A. 2014 Jul;164A(7):1635-41. doi: 10.1002/ajmg.a.36502. Epub 2014 Mar 26. PubMed PMID: 24677493.
25. Mäkitie O, Geiberger S, Horemuzova E, Hagenäs L, Moström E, Nordenskjöld M, **Grigelioniene G**, Nordgren A. *SLC26A2* disease spectrum in Sweden - high frequency of recessive multiple epiphyseal dysplasia (rMED). Clin Genet. 2015 Mar;87(3):273-8. doi: 10.1111/cge.12371. Epub 2014 Apr 1. PubMed PMID: 24598000.
26. Lindstrand A#, **Grigelioniene G#,** Nilsson D, Pettersson M, Hofmeister W, Anderlid BM, Kant SG, Ruivenkamp CA, Gustavsson P, Valta H, Geiberger S, Topa A, Lagerstedt-Robinson K, Taylan F, Wincent J, Laurell T, Pekkinen M, Nordenskjöld M, Mäkitie O, Nordgren A. Different mutations in *PDE4D* associated with developmental disorders with mirror phenotypes. J Med Genet. 2014. Jan;51(1):45-54. doi: 10.1136/jmedgenet-2013-101937. Epub 2013 Nov 7. PubMed PMID: 24203977. # equal contribution.
27. **Grigelioniene G**, Geiberger S, Papadogiannakis N, Mäkitie O, Nishimura G, Nordgren A, Conner P. The phenotype range of achondrogenesis 1A. Am J Med Genet A. 2013 Oct;161A(10):2554-8. doi: 10.1002/ajmg.a.36106. Epub 2013 Aug 16. PubMed PMID: 23956106.
28. Soderhall C, Lundin J, Lagerstedt-Robinson K, **Grigelioniene G**, Lackgren G, Kockum CC, Nordenskjold A. A case with bladder exstrophy and unbalanced X chromosome rearrangement. Eur J Pediatr Surg. 2014 Aug;24(4):353-9. doi: 10.1055/s-0033-1349056. Epub 2013 Jun 25. PubMed PMID: 23801353.
29. Unger S, Górna MW, Le Béchec A, Do Vale-Pereira S, Bedeschi MF, Geiberger S, **Grigelioniene G,** Horemuzova E, Lalatta F, Lausch E, Magnani C, Nampoothiri S, Nishimura G, Petrella D, Rojas-Ringeling F, Utsunomiya A, Zabel B, Pradervand S, Harshman K, Campos-Xavier B, Bonafé L,

Superti-Furga G, Stevenson B, Superti-Furga A. *FAM111A* mutations result in hypoparathyroidism and impaired skeletal development. Am J Hum Genet. 2013 Jun 6;92(6):990-5. doi: 10.1016/j.ajhg.2013.04.020. Epub 2013 May 16. PubMed PMID: 23684011.
30. Laurell T, Lundin J, Anderlid BM, Gorski JL, **Grigelioniene G,** Knight SJ, Krepischi AC, Nordenskjöld A, Price SM, Rosenberg C, Turnpenny PD,
Vianna-Morgante AM, Nordgren A. Molecular and clinical delineation of the 17q22 microdeletion phenotype. Eur J Hum Genet. 2013 Oct;21(10):1085-92. doi:10.1038/ejhg.2012.306. Epub 2013 Jan 30. PubMed PMID: 23361222.
31. Laurell T, Vandermeer JE, Wenger AM, **Grigelioniene G**, Nordenskjöld A, Arner M, Ekblom AG, Bejerano G, Ahituv N, Nordgren A. A novel 13 base pair insertion in the sonic hedgehog ZRS limb enhancer (ZRS/LMBR1) causes preaxial polydactyly with triphalangeal thumb. Hum Mutat. 2012 Jul;33(7):1063-6. doi: 10.1002/humu.22097. Epub 2012 May 11. PubMed PMID: 22495965.
32. **Grigelioniene G**, Papadogiannakis N, Conner P, Geiberger S, Nishikawa M, Nakayama M. Extending the phenotype of lethal skeletal dysplasia type al Gazali. Am J Med Genet A. 2011 Jun;155A(6):1404-8. doi: 10.1002/ajmg.a.33990. Epub 2011 May 12. PubMed PMID: 21567921.

Peer-reviewed conference contributions:
1. Hammarsjö A, Wang Z, Vaz R, Taylan F, Sedghi M, Girisha KM, Chitayat D, Neethukrishna K, Godoy R, Gowrishankar K, Lindstrand A, Nasiri J, Baktashian M, Newton PT, Guo L, Petterson M, Chagin AS, Nishimura G, Yan L, Matsumoto N, Shannon P, Nordgren A, Miyake N, **Grigelioniene G**[*], Ikegawa S[*]. Skeletal dysplasia combined with Joubert syndrome: a novel skeletal ciliopathy phenotype. 13th ISDS meeting Sept 20–23, 2017 Bruges, Belgium. *egual contribution.
2. Leal GF, Nishimura G, Bertola DR, Yamamoto GL, Voss U, Hammarsjö A, Horemuzova E, Papadiogannakis N, Tham E, **Grigelioniene G**. Kyfomelic dysplasia including Kozlowski-Reardon alike phenotype is at the most severe end of lysis-hydroxylase-2-related skeletal disorders. 13th ISDS meeting Sept 20-23, 2017 Bruges, Belgium.
3. Warman M & Grigelioniene G. A neomorphic mutation in a microRNA gene causes a new human skeletal dysplasia (oral presentation). 13th ISDS meeting Sept 20, 2017, Bruges, Belgium.
4. Garza-Flores A, Dubuc A, Dias-Santagata D; Hammarsjö A, **Grigelioniene G**, Lin A, PC. Goldenberg PC. A case of Axenfeld-Rieger syndrome and skeletal dysplasia with a 1.9 kb *FOXC1* deletion: Narrowing the critical region of a previously-described rare phenotype. 37th Annual David W. *Smith Workshop* on Malformations and Morphogenesis, Sept 9–14, 2016 University of California, Los Angeles *Conference* Center, Lake Arrowhead, CA.
5. Hammarsjö A, Taylan F, Pettersson M, Lagerstedt-Robinson K, Malmgren H, Beleza-Meireles A, Girisha K, Shimizu K, Horemuzova E, Sahai I, Traum A, Lin A, Jüppner H, Chitayat D, Nordenskjöld M, Nordgren A, Nishimura G, Lindstrand A, Grigelioniene G
"Skeletal ciliopathies: a molecular study of 17 patients with SRTD and Sensenbrenner syndrome" 64th Annual *Meeting* of the American Society of Human Genetics, Oct 18–22 2014, San Diego Convention Center, San Diego, CA.

Other publications including popular science books/presentations
Achondrogenesis (2016) National Organisation for Rare Disease, USA
https://rarediseases.org/rare-diseases/achondrogenesis/
Recessive multiple epifyseal dysplasia (2015) National Organisation for Rare Disease, USA.
https://rarediseases.org/rare-diseases/recessive-multiple-epiphyseal-dysplasia/
Chondrodysplasia punctata (2015) Socialstyrelsen.
http://www.socialstyrelsen.se/ovanligadiagnoser/chondrodysplasiapunctata

Invited lectures/presentations abroad:
1. Clinical and Molecular Diagnosis of Skeletal Dysplasias. Massachusetts General Hospital, Harvard Medical School, Boston, 20160223.
2. Clinical and Molecular Studies on Congenital Syndromes Involving the Skeleton. Massachusetts General Hospital, Harvard Medical School 20160212.
3. A diagnostic skeletal dysplasia gene panel: the Swedish experience. Scandinavian Skeletal Dysplasia Workshop 2015 Oslo, Norway 20160310.
4. Pseudohypoparathyroidism Ia, 20141121, Metabolic Bone Workshop, Stockholm.
5. Skeletal dysplasia panels and beyond, Nordic Skeletal symposium, Copenhagen, Denmark 20180309.