

# Giedre Grigelioniene

## List of Publications by Year in descending order

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Version: 2024-02-01

65  
papers

2,319  
citations

201658

27  
h-index

233409

45  
g-index

70  
all docs

70  
docs citations

70  
times ranked

3896  
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic epidemiology, prevalence, and genotype-phenotype correlations in the Swedish population with osteogenesis imperfecta. <i>European Journal of Human Genetics</i> , 2015, 23, 1042-1050.	2.8	126
2	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	8.2	116
3	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. <i>American Journal of Human Genetics</i> , 2013, 92, 990-995.	6.2	114
4	Demonstration of Estrogen Receptor- $\beta$ Immunoreactivity in Human Growth Plate Cartilage. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 370-373.	3.6	113
5	Mutations in SPINT2 Cause a Syndromic Form of Congenital Sodium Diarrhea. <i>American Journal of Human Genetics</i> , 2009, 84, 188-196.	6.2	110
6	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. <i>American Journal of Human Genetics</i> , 2014, 95, 285-293.	6.2	110
7	Demonstration of Estrogen Receptor- $\beta$ Immunoreactivity in Human Growth Plate Cartilage. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 370-373.	3.6	99
8	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. <i>Genome Medicine</i> , 2019, 11, 68.	8.2	88
9	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. <i>Nature Medicine</i> , 2019, 25, 583-590.	30.7	86
10	Extending the phenotype of BMPER-related skeletal dysplasias to ischiopspinal dysostosis. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 1.	2.7	70
11	Comprehensive genetic and epigenetic analysis of sporadic meningioma for macro-mutations on 22q and micro-mutations within the NF2 locus. <i>BMC Genomics</i> , 2007, 8, 16.	2.8	67
12	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. <i>European Journal of Human Genetics</i> , 2015, 23, 1176-1185.	2.8	67
13	Mutations in COL1A1 and COL1A2 and dental aberrations in children and adolescents with osteogenesis imperfecta - A retrospective cohort study. <i>PLoS ONE</i> , 2017, 12, e0176466.	2.5	62
14	Analysis of short stature homeobox-containing gene ( SHOX ) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. <i>Human Genetics</i> , 2001, 109, 551-558.	3.8	60
15	Novel and recurrent TRPV4 mutations and their association with distinct phenotypes within the TRPV4 dysplasia family. <i>Journal of Medical Genetics</i> , 2010, 47, 704-709.	3.2	58
16	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. <i>American Journal of Human Genetics</i> , 2018, 103, 553-567.	6.2	58
17	Different mutations in <i>PDE4D</i> associated with developmental disorders with mirror phenotypes. <i>Journal of Medical Genetics</i> , 2014, 51, 45-54.	3.2	57
18	Clinical and molecular characterization of duplications encompassing the human <i>SHOX</i> gene reveal a variable effect on stature. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1407-1414.	1.2	46

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19	Spondyloocular Syndrome: Novel Mutations in <i>XYLT2</i> Gene and Expansion of the Phenotypic Spectrum. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 1577-1585.	2.8	43
20	Growth in achondroplasia: Development of height, weight, head circumference, and body mass index in a European cohort. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1723-1734.	1.2	42
21	Absence of GP130 cytokine receptor signaling causes extended StÅ¼ve-Wiedemann syndrome. <i>Journal of Experimental Medicine</i> , 2020, 217, .	8.5	41
22	Mutations in short stature homeobox containing gene ( <i>SHOX</i> ) in dyschondrosteosis but not in hypochondroplasia. <i>Human Genetics</i> , 2000, 107, 145-149.	3.8	37
23	Tooth agenesis in osteogenesis imperfecta related to mutations in the collagen type I genes. <i>Oral Diseases</i> , 2017, 23, 42-49.	3.0	36
24	Decreased fracture rate, pharmacogenetics and BMD response in 79 Swedish children with osteogenesis imperfecta types I, III and IV treated with Pamidronate. <i>Bone</i> , 2016, 87, 11-18.	2.9	33
25	Axial Spondylometaphyseal Dysplasia Is Caused by <i>C21orf2</i> Mutations. <i>PLoS ONE</i> , 2016, 11, e0150555.	2.5	32
26	Analysis of Multiple Families With Single Individuals Affected by Pseudohypoparathyroidism Type Ib (PHP1B) Reveals Only One Novel Maternally Inherited <i>GNAS</i> Deletion. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 796-805.	2.8	31
27	Skeletal ciliopathies: a pattern recognition approach. <i>Japanese Journal of Radiology</i> , 2020, 38, 193-206.	2.4	30
28	A novel 13 base pair insertion in the sonic hedgehog ZRS limb enhancer ( <i>ZRS/LMBR1</i> ) causes preaxial polydactyly with triphalangeal thumb. <i>Human Mutation</i> , 2012, 33, 1063-1066.	2.5	29
29	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€™Nishimura skeletal dysplasia due to pathogenic variants in <i>ALG9</i> . <i>European Journal of Human Genetics</i> , 2016, 24, 198-207.	2.8	29
30	Development of body proportions in achondroplasia: Sitting height, leg length, arm span, and foot length. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1819-1829.	1.2	26
31	Estrogens and human growth. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2000, 74, 383-386.	2.5	25
32	Axial spondylometaphyseal dysplasia is also caused by <i>NEK1</i> mutations. <i>Journal of Human Genetics</i> , 2017, 62, 503-506.	2.3	25
33	Short Stature in KBC Syndrome: First Responses to Growth Hormone Treatment. <i>Hormone Research in Paediatrics</i> , 2015, 83, 361-364.	1.8	24
34	A Large Inversion Involving <i>GNAS</i> Exon A/B and All Exons Encoding <i>GsÎ±</i> Is Associated With Autosomal Dominant Pseudohypoparathyroidism Type Ib (PHP1B). <i>Journal of Bone and Mineral Research</i> , 2017, 32, 776-783.	2.8	22
35	Novel <i>KIAA0753</i> mutations extend the phenotype of skeletal ciliopathies. <i>Scientific Reports</i> , 2017, 7, 15585.	3.3	21
36	Expanding the Clinical Spectrum of Phenotypes Caused by Pathogenic Variants in <i>PLOD2</i> . <i>Journal of Bone and Mineral Research</i> , 2018, 33, 753-760.	2.8	20

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37	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. <i>Journal of Human Genetics</i> , 2021, 66, 995-1008.	2.3	19
38	Deconvolution of seed and RNA-binding protein crosstalk in RNAi-based functional genomics. <i>Nature Genetics</i> , 2018, 50, 657-661.	21.4	18
39	Identification of three novel <i>FGF16</i> mutations in X-linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2014, 2, 402-411.	1.2	17
40	Mesomelic and rhizomelic short stature: The phenotype of combined Leri-Weill dyschondrosteosis and achondroplasia or hypochondroplasia. <i>American Journal of Medical Genetics Part A</i> , 2003, 116A, 61-65.	2.4	16
41	<i>Alu-Alu</i> mediated intragenic duplications in <i>IFT81</i> and <i>MATN3</i> are associated with skeletal dysplasias. <i>Human Mutation</i> , 2018, 39, 1456-1467.	2.5	16
42	Cerebrospinal Fluid of Newborn Infants Contains a Deglycosylated Form of the Intermediate Filament Nestin. <i>Pediatric Research</i> , 1996, 40, 809-814.	2.3	15
43	<i>SLC26A2</i> disease spectrum in Sweden—high frequency of recessive multiple epiphyseal dysplasia ( <i>rMED</i> ). <i>Clinical Genetics</i> , 2015, 87, 273-278.	2.0	13
44	Novel mutations in the <i>LRP5</i> gene in patients with Osteoporosis-pseudoglioma syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3132-3135.	1.2	13
45	<i>SLC4A2</i> Deficiency Causes a New Type of Osteopetrosis. <i>Journal of Bone and Mineral Research</i> , 2020, 37, 226-235.	2.8	12
46	Molecular and clinical delineation of the 17q22 microdeletion phenotype. <i>European Journal of Human Genetics</i> , 2013, 21, 1085-1092.	2.8	11
47	Defining the clinical phenotype of Saul-Wilson syndrome. <i>Genetics in Medicine</i> , 2020, 22, 857-866.	2.4	11
48	Autosomal recessive mutations in the <i>COL2A1</i> gene cause severe spondyloepiphyseal dysplasia. <i>Clinical Genetics</i> , 2015, 87, 496-498.	2.0	9
49	Asn540Lys mutation in fibroblast growth factor receptor 3 and phenotype in hypochondroplasia. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2000, 89, 1072-1076.	1.5	9
50	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 11.	3.8	7
51	Autosomal dominant brachyolmia in a large Swedish family: Phenotypic spectrum and natural course. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1635-1641.	1.2	6
52	A Case with Bladder Exstrophy and Unbalanced X Chromosome Rearrangement. <i>European Journal of Pediatric Surgery</i> , 2014, 24, 353-359.	1.3	5
53	Radiologic Features of Type II and Type XI Collagenopathies. <i>Radiographics</i> , 2021, 41, 192-209.	3.3	5
54	Lack of <i>GNAS</i> Remethylation During Oogenesis May Be a Cause of Sporadic Pseudohypoparathyroidism Type Ib. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e1610-e1619.	3.6	5

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55	Metacarpophalangeal pattern profile analysis in Ieriâ€weill dyschondrosteosis. Acta Radiologica, 2005, 46, 200-207.	1.1	4
56	The phenotype range of achondrogenesis 1A. , 2013, 161A, n/a-n/a.		4
57	Autosomal recessive brachyolmia: early radiological findings. Skeletal Radiology, 2016, 45, 1557-1560.	2.0	4
58	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for nonâ€oncologic disorders. American Journal of Medical Genetics, Part A, 2021, 185, 517-527.	1.2	3
59	Pathogenic variant in the <i>COL2A1</i> gene is associated with Spondyloepiphyseal dysplasia type Stanescu. American Journal of Medical Genetics, Part A, 2016, 170, 266-269.	1.2	2
60	Genotype-Phenotype Correlation of PLOD2 Skeletal Dysplasias Using Structural Information. Journal of Bone and Mineral Research, 2018, 33, 1377-1378.	2.8	2
61	A novel missense mutation Ile538Val in the fibroblast growth factor receptor 3 in hypochondroplasia. Human Mutation, 1998, 11, 333-333.	2.5	2
62	Extending the phenotype of lethal skeletal dysplasia type al Gazali. American Journal of Medical Genetics, Part A, 2011, 155, 1404-1408.	1.2	1
63	SAT0493â€...Farber Disease: First Natural History Cohort Demonstrates a Broad Clinical Spectrum with Implications for Juvenile Idiopathic Arthritis Patients. Annals of the Rheumatic Diseases, 2015, 74, 838.3-839.	0.9	1
64	Exploring human genetic skeletal disorders provides important insights into skeletogenesis and elucidates basic developmental signaling pathways. EBioMedicine, 2020, 62, 103091.	6.1	0
65	Genetics of Achondroplasia and Hypochondroplasia. , 2004, , 349-359.		0