Anja L Frederiksen

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6197469/publications.pdf

Version: 2024-02-01

36	671	706676 14	651938 25
papers	citations	h-index	g-index

36	36	36	1148
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Bone-microarchitecture and bone-strength in a sample of adults with hypophosphatasia and a matched reference population assessed by HR-pQCT and impact microindentation. Bone, 2022, 160, 116420.	1.4	3
2	PHKA2 variants expand the phenotype of phosphorylase B kinase deficiency to include patients with ketotic hypoglycemia only. American Journal of Medical Genetics, Part A, 2021, 185, 2959-2975.	0.7	2
3	Biochemical, clinical and genetic characteristics in adults with persistent hypophosphatasaemia; Data from an endocrinological outpatient clinic in Denmark. Bone Reports, 2021, 15, 101101.	0.2	4
4	Modeling-based bone formation transforms trabeculae to cortical bone in the sclerotic areas in Buschke-Ollendorff syndrome. A case study of two females with LEMD3 variants. Bone, 2020, 135, 115313.	1.4	6
5	Variability in Medullary Thyroid Carcinoma in RET L790F Carriers: A Case Comparison Study of Index Patients. Frontiers in Endocrinology, 2020, 11, 251.	1.5	5
6	Clinical Features of Multiple Endocrine Neoplasia Type 4: Novel Pathogenic Variant and Review of Published Cases. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3637-3646.	1.8	83
7	Multiple Fractures and Impaired Bone Fracture Healing in a Patient with Pycnodysostosis and Hypophosphatasia. Calcified Tissue International, 2019, 105, 681-686.	1.5	5
8	Absence of an osteopetrosis phenotype in IKBKG (NEMO) mutation-positive women: A case-control study. Bone, 2019, 121, 243-254.	1.4	4
9	Completeness of RET testing in patients with medullary thyroid carcinoma in Denmark 1997–2013: a nationwide study. Clinical Epidemiology, 2019, Volume 11, 93-99.	1.5	7
10	Survival and Long-Term Biochemical Cure in Medullary Thyroid Carcinoma in Denmark 1997â€“2014: A Nationwide Study. Thyroid, 2019, 29, 368-377.	2.4	43
11	Mitochondrial mutation m.3243A>G associates with insulin resistance in non-diabetic carriers. Endocrine Connections, 2019, 8, 829-837.	0.8	4
12	Lecocytes mutation load declines with age in carriers of the m.3243A>G mutation: A 10â€year Prospective Cohort. Clinical Genetics, 2018, 93, 925-928.	1.0	11
13	Incidence and prevalence of multiple endocrine neoplasia 2A in Denmark 1901–2014: a nationwide study. Clinical Epidemiology, 2018, Volume 10, 1479-1487.	1.5	19
14	Incidence and prevalence of sporadic and hereditary MTC in Denmark 1960â€“2014: a nationwide study. Endocrine Connections, 2018, 7, 829-839.	0.8	32
15	Incidence and prevalence of multiple endocrine neoplasia 2B in Denmark: a nationwide study. Endocrine-Related Cancer, 2017, 24, L39-L42.	1.6	32
16	Novel Somatic RET Mutation Questioning the Causality of the RET I852M Germline Sequence Variant in Multiple Endocrine Neoplasia 2A. Thyroid, 2017, 27, 1103-1104.	2.4	13
17	Mitochondrial Point Mutation m.3243A>G Associates With Lower Bone Mineral Density, Thinner Cortices, and Reduced Bone Strength: A Case-Control Study. Journal of Bone and Mineral Research, 2017, 32, 2041-2048.	3.1	9
18	Founder Effect of the RET^{C611Y} Mutation in Multiple Endocrine Neoplasia 2A in Denmark: A Nationwide Study. Thyroid, 2017, 27, 1505-1510.	2.4	16

#	Article	IF	Citations
19	Reply to: Reduced Bone Mineral Density in m.3243A>G Carriers May Be Multifactorial. Journal of Bone and Mineral Research, 2017, 32, 2317-2318.	3.1	0
20	Distribution of RET Mutations in Multiple Endocrine Neoplasia 2 in Denmark 1994â€“2014: A Nationwide Study. Thyroid, 2017, 27, 215-223.	2.4	29
21	Boucher NeuhÃuser Syndrome â€“ A rare cause of inherited hypogonadotropic hypogonadism. A case of two adult siblings with two novel mutations in PNPLA6. European Journal of Medical Genetics, 2017, 60, 105-109.	0.7	15
22	Systematic genetic screening in a prospective group of Danish patients with pheochromocytoma. Research and Reports in Urology, 2017, Volume 9, 113-119.	0.6	0
23	Octreotide therapy and restricted fetal growth: pregnancy in familial hyperinsulinemic hypoglycemia. Endocrinology, Diabetes and Metabolism Case Reports, 2017, 2017, .	0.2	6
24	Asymptomatic parental mosaicism for osteogenesis imperfecta associated with a new splice site mutation in COL 1A2. Clinical Case Reports (discontinued), 2016, 4, 972-978.	0.2	4
25	Bone structure in two adult subjects with impaired minor spliceosome function resulting from RNU4ATAC mutations causing microcephalic osteodysplastic primordial dwarfism type 1 (MOPD1). Bone, 2016, 92, 145-149.	1.4	8
26	Prevalence of migraine in persons with the 3243A>G mutation in mitochondrial <scp>DNA</scp>. European Journal of Neurology, 2016, 23, 175-181.	1.7	31
27	MP12-01 SYSTEMATIC GENETIC SCREENING IN A PROSPECTIVE GROUP OF DANISH PATIENTS WITH PHEOCHROMOCYTOMA. Journal of Urology, 2016, 195, .	0.2	0
28	Neonatal High Bone Mass With First Mutation of the NF-ÎºB Complex: Heterozygous De Novo Missense (p.Asp512Ser) RELA (Rela/p65). Journal of Bone and Mineral Research, 2016, 31, 163-172.	3.1	21
29	Case report: vitamin Dâ€dependent rickets type 1 caused by a novel <scp>CYP</scp>27B1 mutation. Clinical Case Reports (discontinued), 2015, 3, 1012-1016.	0.2	9
30	Increased cortical area and thickness in the distal radius in subjects with SHOX-gene mutation. Bone, 2014, 69, 23-29.	1.4	15
31	Blue cone monochromatism in a female due to skewed X-inactivation. Ophthalmic Genetics, 2013, 34, 101-104.	0.5	4
32	A novel mitochondrial mutation m.8989G>C associated with neuropathy, ataxia, retinitis pigmentosa â€" The NARP syndrome. Gene, 2013, 515, 372-375.	1.0	35
33	Limited diagnostic value of enzyme analysis in patients with mitochondrial tRNA mutations. Muscle and Nerve, 2010, 41, 607-613.	1.0	20
34	High Prevalence of Impaired Glucose Homeostasis and Myopathy in Asymptomatic and Oligosymptomatic 3243A>G Mitochondrial DNA Mutation-Positive Subjects. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 2872-2879.	1.8	18
35	Muscle Phenotype and Mutation Load in 51 Persons With the 3243A>G Mitochondrial DNA Mutation. Archives of Neurology, 2006, 63, 1701.	4.9	71
36	Tissue specific distribution of the 3243A->G mtDNA mutation. Journal of Medical Genetics, 2006, 43, 671-677.	1.5	87