

Joakim Klar

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9185321/publications.pdf>

Version: 2024-02-01

71
papers

3,503
citations

201385

27
h-index

138251

58
g-index

71
all docs

71
docs citations

71
times ranked

6132
citing authors

#	ARTICLE	IF	CITATIONS
1	The gene encoding ribosomal protein S19 is mutated in Diamond-Blackfan anaemia. <i>Nature Genetics</i> , 1999, 21, 169-175.	9.4	747
2	Vascular endothelial growth factor B controls endothelial fatty acid uptake. <i>Nature</i> , 2010, 464, 917-921.	13.7	423
3	Vascular Endothelial Growth Factor-B Induces Myocardium-Specific Angiogenesis and Arteriogenesis via Vascular Endothelial Growth Factor Receptor-1 and Neuropilin Receptor-1 Dependent Mechanisms. <i>Circulation</i> , 2009, 119, 845-856.	1.6	172
4	Mutations in the Fatty Acid Transport Protein 4 Gene Cause the Ichthyosis Prematurity Syndrome. <i>American Journal of Human Genetics</i> , 2009, 85, 248-253.	2.6	147
5	Mutations in the gene encoding fibroblast growth factor 10 are associated with aplasia of lacrimal and salivary glands. <i>Nature Genetics</i> , 2005, 37, 125-128.	9.4	144
6	Alpha-cardiac actin mutations produce atrial septal defects. <i>Human Molecular Genetics</i> , 2008, 17, 256-265.	1.4	128
7	Isolated oligodontia associated with mutations in <i>EDARADD</i> , <i>AXIN2</i> , <i>MSX1</i> , and <i>PAX9</i> genes. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1616-1622.	0.7	124
8	Welander Distal Myopathy Caused by an Ancient Founder Mutation in <i>TIA1</i> Associated with Perturbed Splicing. <i>Human Mutation</i> , 2013, 34, n/a-n/a.	1.1	91
9	A Single-Nucleotide Deletion in the POMP 5' UTR Causes a Transcriptional Switch and Altered Epidermal Proteasome Distribution in KLICK Genodermatosis. <i>American Journal of Human Genetics</i> , 2010, 86, 596-603.	2.6	79
10	Variant Phenotype of Best Vitelliform Macular Dystrophy Associated with Compound Heterozygous Mutations in <i>VMD2</i> . <i>Ophthalmic Genetics</i> , 2006, 27, 51-56.	0.5	74
11	<i>WNT10A</i> mutations account for 1/4 of population-based isolated oligodontia and show phenotypic correlations. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 353-359.	0.7	69
12	<i>WNT10A</i> missense mutation associated with a complete Odonto-Onycho-Dermal Dysplasia syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 1600-1605.	1.4	64
13	Abolished <i>InsP3R2</i> function inhibits sweat secretion in both humans and mice. <i>Journal of Clinical Investigation</i> , 2014, 124, 4773-4780.	3.9	63
14	Congenital ichthyosis: mutations in <i>ichthyin</i> are associated with specific structural abnormalities in the granular layer of epidermis. <i>Journal of Medical Genetics</i> , 2007, 44, 615-620.	1.5	59
15	Mutations in <i>Frizzled 6</i> Cause Isolated Autosomal-Recessive Nail Dysplasia. <i>American Journal of Human Genetics</i> , 2011, 88, 852-860.	2.6	58
16	A Meniere's disease gene linked to chromosome 12p12.3. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2006, 141B, 463-467.	1.1	56
17	Fibroblast growth factor 10 haploinsufficiency causes chronic obstructive pulmonary disease. <i>Journal of Medical Genetics</i> , 2011, 48, 705-709.	1.5	54
18	Altered paracellular cation permeability due to a rare <i>CLDN10B</i> variant causes anhidrosis and kidney damage. <i>PLoS Genetics</i> , 2017, 13, e1006897.	1.5	50

#	ARTICLE	IF	CITATIONS
19	SNX10 gene mutation leading to osteopetrosis with dysfunctional osteoclasts. <i>Scientific Reports</i> , 2017, 7, 3012.	1.6	43
20	Familial Ménière's Disease in Five Generations. <i>Otology and Neurotology</i> , 2006, 27, 681-686.	0.7	42
21	<i>muSK</i> : a new target for lethal fetal akinesia deformation sequence (FADS). <i>Journal of Medical Genetics</i> , 2015, 52, 195-202.	1.5	41
22	Truncating ribosomal protein S19 mutations and variable clinical expression in Diamond-Blackfan anemia. <i>Human Genetics</i> , 1999, 105, 496-500.	1.8	40
23	Recurrent <i>GATA1</i> mutations in Diamond-Blackfan anaemia. <i>British Journal of Haematology</i> , 2014, 166, 949-951.	1.2	40
24	Transcriptome and Proteome Profiling of Neural Induced Pluripotent Stem Cells from Individuals with Down Syndrome Disclose Dynamic Dysregulations of Key Pathways and Cellular Functions. <i>Molecular Neurobiology</i> , 2019, 56, 7113-7127.	1.9	36
25	Evidence for autosomal recessive inheritance in SPG3A caused by homozygosity for a novel <i>ATL1</i> missense mutation. <i>European Journal of Human Genetics</i> , 2014, 22, 1180-1184.	1.4	35
26	Transcriptomes of Dravet syndrome iPSC derived GABAergic cells reveal dysregulated pathways for chromatin remodeling and neurodevelopment. <i>Neurobiology of Disease</i> , 2019, 132, 104583.	2.1	32
27	<i>Frizzled6</i> Deficiency Disrupts the Differentiation Process of Nail Development. <i>Journal of Investigative Dermatology</i> , 2013, 133, 1990-1997.	0.3	30
28	<i>FATP4</i> missense and nonsense mutations cause similar features in Ichthyosis Prematurity Syndrome. <i>BMC Research Notes</i> , 2011, 4, 90.	0.6	27
29	Phenotypic expansion of visceral myopathy associated with <i>ACTG2</i> tandem base substitution. <i>European Journal of Human Genetics</i> , 2015, 23, 1679-1683.	1.4	27
30	A novel variant in <i>MYLK</i> causes thoracic aortic dissections: genotypic and phenotypic description. <i>BMC Medical Genetics</i> , 2016, 17, 61.	2.1	27
31	A missense variant in <i>ITPR1</i> provides evidence for autosomal recessive SCA29 with asymptomatic cerebellar hypoplasia in carriers. <i>European Journal of Human Genetics</i> , 2017, 25, 848-853.	1.4	27
32	Assignment of the locus for ichthyosis prematurity syndrome to chromosome 9q33.3-34.13. <i>Journal of Medical Genetics</i> , 2004, 41, 208-212.	1.5	26
33	Familial Ménière's disease restricted to 1.48 Mb on chromosome 12p12.3 by allelic and haplotype association. <i>Journal of Human Genetics</i> , 2010, 55, 834-837.	1.1	26
34	Cenani's Lenz syndrome restricted to limb and kidney anomalies associated with a novel <i>LRP4</i> missense mutation. <i>European Journal of Medical Genetics</i> , 2013, 56, 371-374.	0.7	22
35	A novel <i>AP4M1</i> mutation in autosomal recessive cerebral palsy syndrome and clinical expansion of <i>AP-4</i> deficiency. <i>BMC Medical Genetics</i> , 2014, 15, 133.	2.1	22
36	Homozygous <i>GRID2</i> missense mutation predicts a shift in the D-serine binding domain of <i>GluD2</i> in a case with generalized brain atrophy and unusual clinical features. <i>BMC Medical Genetics</i> , 2017, 18, 144.	2.1	21

#	ARTICLE	IF	CITATIONS
37	A <i>BBS1</i> SVA retrotransposon insertion is a frequent cause of Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2021, 99, 318-324.	1.0	21
38	Truncating ribosomal protein S19 mutations and variable clinical expression in Diamond-Blackfan anemia. <i>Human Genetics</i> , 1999, 105, 496-500.	1.8	20
39	DNA methylation changes in Down syndrome derived neural iPSCs uncover co-dysregulation of ZNF and HOX3 families of transcription factors. <i>Clinical Epigenetics</i> , 2020, 12, 9.	1.8	20
40	Novel SACS mutations associated with intellectual disability, epilepsy and widespread supratentorial abnormalities. <i>Journal of the Neurological Sciences</i> , 2016, 371, 105-111.	0.3	19
41	A founder mutation for ichthyosis prematurity syndrome restricted to 76 kb by haplotype association. <i>Journal of Human Genetics</i> , 2006, 51, 864-871.	1.1	18
42	Exome sequencing circumvents missing clinical data and identifies a BSCL2 mutation in congenital lipodystrophy. <i>BMC Medical Genetics</i> , 2014, 15, 71.	2.1	17
43	Abnormal primary and permanent dentitions with ectodermal symptoms predict WNT10A deficiency. <i>BMC Medical Genetics</i> , 2016, 17, 88.	2.1	17
44	Novel missense mutation in the RSPO4 gene in congenital hyponychia and evidence for a polymorphic initiation codon (p.M1). <i>BMC Medical Genetics</i> , 2012, 13, 120.	2.1	16
45	Autosomal Recessive Transmission of a Rare KRT74 Variant Causes Hair and Nail Ectodermal Dysplasia: Allelism with Dominant Woolly Hair/Hypotrichosis. <i>PLoS ONE</i> , 2014, 9, e93607.	1.1	16
46	Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy. <i>American Journal of Human Genetics</i> , 2021, 108, 739-748.	2.6	15
47	RAR-related orphan receptor A isoform 1 (RORa1) is disrupted by a balanced translocation t(4;15)(q22.3;q21.3) associated with severe obesity. <i>European Journal of Human Genetics</i> , 2005, 13, 928-934.	1.4	14
48	Whole exome sequencing identifies <i>LRP1</i> as a pathogenic gene in autosomal recessive keratosis pilaris atrophicans. <i>Journal of Medical Genetics</i> , 2015, 52, 599-606.	1.5	14
49	A chromosome 10 variant with a 12 Mb inversion [inv(10)(q11.22q21.1)] identical by descent and frequent in the Swedish population. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 380-386.	0.7	13
50	Familial transient erythroblastopenia of childhood is associated with the chromosome 19q13.2 region but not caused by mutations in coding sequences of the ribosomal protein S19 (RPS19) gene. <i>British Journal of Haematology</i> , 2002, 119, 261-264.	1.2	12
51	Targeted high-throughput sequencing of candidate genes for chronic obstructive pulmonary disease. <i>BMC Pulmonary Medicine</i> , 2016, 16, 146.	0.8	12
52	Stereocilin gene variants associated with episodic vertigo: expansion of the DFNB16 phenotype. <i>European Journal of Human Genetics</i> , 2018, 26, 1871-1874.	1.4	12
53	Phenotypic variability in a seven-generation Swedish family segregating autosomal dominant hearing impairment due to a novel EYA4 frameshift mutation. <i>Gene</i> , 2015, 563, 10-16.	1.0	11
54	Autosomal recessive pure hair and nail ectodermal dysplasia linked to chromosome 12p11.1-q14.3 without KRTHB5 gene mutation. <i>European Journal of Dermatology</i> , 2010, 20, 443-446.	0.3	10

#	ARTICLE	IF	CITATIONS
55	A novel mutation in the Lipase H gene underlies autosomal recessive hypotrichosis and woolly hair. <i>Scientific Reports</i> , 2012, 2, 730.	1.6	10
56	Cooperative effect of ribosomal protein s19 and Pim-1 kinase on murine c-Myc expression and myeloid/erythroid cellularity. <i>Journal of Molecular Medicine</i> , 2010, 88, 39-46.	1.7	8
57	Whole exome sequencing identifies novel variant underlying hereditary spastic paraplegia in consanguineous Pakistani families. <i>Journal of Clinical Neuroscience</i> , 2019, 67, 19-23.	0.8	8
58	New perspectives on the dynamic behaviour of oral lichen planus. <i>European Journal of Dermatology</i> , 2012, 22, 178-181.	0.3	6
59	Homozygosity for a missense variant in <i>COMP</i> gene associated with severe pseudoachondroplasia. <i>Clinical Genetics</i> , 2018, 93, 182-186.	1.0	5
60	Aberrant splicing due to a novel RPS7 variant causes Diamond-Blackfan Anemia associated with spontaneous remission and meningocele. <i>International Journal of Hematology</i> , 2020, 112, 894-899.	0.7	5
61	Expanding the phenotypic spectrum of osteogenesis imperfecta type V including heterotopic ossification of muscle origins and attachments. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00723.	0.6	4
62	Cohort profile: the Swedish study of Sudden cardiac Death in the Young (SUDDY) 2000-2010: a complete nationwide cohort of SCDs. <i>BMJ Open</i> , 2022, 12, e055557.	0.8	4
63	Generation of human induced pluripotent stem cell (iPSC) lines from three patients with von Hippel-Lindau syndrome carrying distinct VHL gene mutations. <i>Stem Cell Research</i> , 2019, 38, 101474.	0.3	3
64	A novel mutation in Lysophosphatidic Acid Receptor 6 gene in autosomal recessive hypotrichosis and evidence for a founder effect. <i>European Journal of Dermatology</i> , 2012, 22, 464-466.	0.3	2
65	Primary microcephaly, primordial dwarfism, and brachydactyly in adult cases with biallelic skipping of <i>RTTN</i> exon 42. <i>Human Mutation</i> , 2019, 40, 899-903.	1.1	2
66	Whole genome sequencing of familial isolated oesophagus atresia uncover shared structural variants. <i>BMC Medical Genomics</i> , 2020, 13, 85.	0.7	2
67	Sudden cardiac death due to ARVC in the young: molecular autopsy by whole exome sequencing of DNA from dried blood spots (DBS) collected at birth. <i>Europace</i> , 2021, 23, .	0.7	1
68	Cooperative effect of ribosomal protein s19 and Pim-1 kinase on murine c-Myc expression and myeloid/erythroid cellularity. <i>Nature Precedings</i> , 2009, , .	0.1	0
69	A Single-Nucleotide Deletion in the POMP 5' UTR Causes a Transcriptional Switch and Altered Epidermal Proteasome Distribution in KLICK Genodermatosis. <i>American Journal of Human Genetics</i> , 2010, 86, 655.	2.6	0
70	Combined Disruptions of the Ribosomal Protein s19 and Pim1 Kinase Genes Are Associated with Increased Myeloid/Erythroid Cellularity and Reduced Apoptosis. <i>Blood</i> , 2008, 112, 3097-3097.	0.6	0
71	Generation of a human iPSC line (UUIGPi015-A) from a patient with Dravet syndrome and a 2.9 Mb deletion spanning SCN1A on chromosome 2. <i>Stem Cell Research</i> , 2022, 60, 102712.	0.3	0