Joakim Klar

List of Publications by Year
in descending order

71	3,503	201385 27	138251 58
papers	citations	h-index	g-index

71	71	71	6132
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Generation of a human iPSC line (UUIGPi015-A) from a patient with Dravet syndrome and a 2.9ÂMb deletion spanning SCN1A on chromosome 2. Stem Cell Research, 2022, 60, 102712.	0.3	0
2	Cohort profile: the Swedish study of SUDden cardiac Death in the Young (SUDDY) 2000â€“2010: a complete nationwide cohort of SCDs. BMJ Open, 2022, 12, e055557.	0.8	4
3	A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324.	1.0	21
4	Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 739-748.	2.6	15
5	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. Europace, 2021, 23, .	0.7	1
6	DNA methylation changes in Down syndrome derived neural iPSCs uncover co-dysregulation of ZNF and HOX3 families of transcription factors. Clinical Epigenetics, 2020, 12, 9.	1.8	20
7	Aberrant splicing due to a novel RPS7 variant causes Diamond-Blackfan Anemia associated with spontaneous remission and meningocele. International Journal of Hematology, 2020, 112, 894-899.	0.7	5
8	Whole genome sequencing of familial isolated oesophagus atresia uncover shared structural variants. BMC Medical Genomics, 2020, 13, 85.	0.7	2
9	Whole exome sequencing identifies novel variant underlying hereditary spastic paraplegia in consanguineous Pakistani families. Journal of Clinical Neuroscience, 2019, 67, 19-23.	0.8	8
10	Transcriptomes of Dravet syndrome iPSC derived GABAergic cells reveal dysregulated pathways for chromatin remodeling and neurodevelopment. Neurobiology of Disease, 2019, 132, 104583.	2.1	32
11	Generation of human induced pluripotent stem cell (iPSC) lines from three patients with von Hippel-Lindau syndrome carrying distinct VHL gene mutations. Stem Cell Research, 2019, 38, 101474.	0.3	3
12	Expanding the phenotypic spectrum of osteogenesis imperfecta type V including heterotopic ossification of muscle origins and attachments. Molecular Genetics & Genomic Medicine, 2019, 7, e00723.	0.6	4
13	Transcriptome and Proteome Profiling of Neural Induced Pluripotent Stem Cells from Individuals with Down Syndrome Disclose Dynamic Dysregulations of Key Pathways and Cellular Functions. Molecular Neurobiology, 2019, 56, 7113-7127.	1.9	36
14	Primary microcephaly, primordial dwarfism, and brachydactyly in adult cases with biallelic skipping of RTTN exon 42. Human Mutation, 2019, 40, 899-903.	1.1	2
15	Homozygosity for a missense variant in <scp>COMP</scp> gene associated with severe pseudoachondroplasia. Clinical Genetics, 2018, 93, 182-186.	1.0	5
16	Stereocilin gene variants associated with episodic vertigo: expansion of the DFNB16 phenotype. European Journal of Human Genetics, 2018, 26, 1871-1874.	1.4	12
17	A missense variant in ITPR1 provides evidence for autosomal recessive SCA29 with asymptomatic cerebellar hypoplasia in carriers. European Journal of Human Genetics, 2017, 25, 848-853.	1.4	27
18	SNX10 gene mutation leading to osteopetrosis with dysfunctional osteoclasts. Scientific Reports, 2017, 7, 3012.	1.6	43

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19	Altered paracellular cation permeability due to a rare CLDN10B variant causes anhidrosis and kidney damage. PLoS Genetics, 2017, 13, e1006897.	1.5	50
20	Homozygous GRID2 missense mutation predicts a shift in the D-serine binding domain of GluD2 in a case with generalized brain atrophy and unusual clinical features. BMC Medical Genetics, 2017, 18, 144.	2.1	21
21	Abnormal primary and permanent dentitions with ectodermal symptoms predict WNT10A deficiency. BMC Medical Genetics, 2016, 17, 88.	2.1	17
22	Novel SACS mutations associated with intellectual disability, epilepsy and widespread supratentorial abnormalities. Journal of the Neurological Sciences, 2016, 371, 105-111.	0.3	19
23	A novel variant in MYLK causes thoracic aortic dissections: genotypic and phenotypic description. BMC Medical Genetics, 2016, 17, 61.	2.1	27
24	Targeted high-throughput sequencing of candidate genes for chronic obstructive pulmonary disease. BMC Pulmonary Medicine, 2016, 16, 146.	0.8	12
25	MuSK: a new target for lethal fetal akinesia deformation sequence (FADS). Journal of Medical Genetics, 2015, 52, 195-202.	1.5	41
26	Whole exome sequencing identifiesLRP1as a pathogenic gene in autosomal recessive keratosis pilaris atrophicans. Journal of Medical Genetics, 2015, 52, 599-606.	1.5	14
27	Phenotypic variability in a seven-generation Swedish family segregating autosomal dominant hearing impairment due to a novel EYA4 frameshift mutation. Gene, 2015, 563, 10-16.	1.0	11
28	Phenotypic expansion of visceral myopathy associated with ACTG2 tandem base substitution. European Journal of Human Genetics, 2015, 23, 1679-1683.	1.4	27
29	A novel AP4M1 mutation in autosomal recessive cerebral palsy syndrome and clinical expansion of AP-4 deficiency. BMC Medical Genetics, 2014, 15, 133.	2.1	22
30	WNT10A mutations account for Â¼ of populationâ€based isolated oligodontia and show phenotypic correlations. American Journal of Medical Genetics, Part A, 2014, 164, 353-359.	0.7	69
31	Recurrent <scp>GATA</scp>1 mutations in <scp>D</scp>iamondâ€<scp>B</scp>lackfan anaemia. British Journal of Haematology, 2014, 166, 949-951.	1.2	40
32	Evidence for autosomal recessive inheritance in SPG3A caused by homozygosity for a novel ATL1 missense mutation. European Journal of Human Genetics, 2014, 22, 1180-1184.	1.4	35
33	Exome sequencing circumvents missing clinical data and identifies a BSCL2 mutation in congenital lipodystrophy. BMC Medical Genetics, 2014, 15, 71.	2.1	17
34	Abolished InsP3R2 function inhibits sweat secretion in both humans and mice. Journal of Clinical Investigation, 2014, 124, 4773-4780.	3.9	63
35	Autosomal Recessive Transmission of a Rare KRT74 Variant Causes Hair and Nail Ectodermal Dysplasia: Allelism with Dominant Woolly Hair/Hypotrichosis. PLoS ONE, 2014, 9, e93607.	1.1	16
36	Welander Distal Myopathy Caused by an Ancient Founder Mutation inTIA1Associated with Perturbed Splicing. Human Mutation, 2013, 34, n/a-n/a.	1.1	91

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37	Cenaniâ€“Lenz syndrome restricted to limb and kidney anomalies associated with a novel LRP4 missense mutation. European Journal of Medical Genetics, 2013, 56, 371-374.	0.7	22
38	Frizzled6 Deficiency Disrupts the Differentiation Process of Nail Development. Journal of Investigative Dermatology, 2013, 133, 1990-1997.	0.3	30
39	A novel mutation in the Lipase H gene underlies autosomal recessive hypotrichosis and woolly hair. Scientific Reports, 2012, 2, 730.	1.6	10
40	A novel mutation in Lysophosphatidic Acid ReceptorÂ6 gene in autosomal recessive hypotrichosis and evidence for a founder effect. European Journal of Dermatology, 2012, 22, 464-466.	0.3	2
41	Novel missense mutation in the RSPO4 gene in congenital hyponychia and evidence for a polymorphic initiation codon (p.M1I). BMC Medical Genetics, 2012, 13, 120.	2.1	16
42	New perspectives on the dynamic behaviour of oral lichen planus. European Journal of Dermatology, 2012, 22, 178-181.	0.3	6
43	FATP4 missense and nonsense mutations cause similar features in Ichthyosis Prematurity Syndrome. BMC Research Notes, 2011, 4, 90.	0.6	27
44	Mutations in Frizzled 6 Cause Isolated Autosomal-Recessive Nail Dysplasia. American Journal of Human Genetics, 2011, 88, 852-860.	2.6	58
45	Isolated oligodontia associated with mutations in EDARADD, AXIN2, MSX1, and PAX9 genes. American Journal of Medical Genetics, Part A, 2011, 155, 1616-1622.	0.7	124
46	Fibroblast growth factor 10 haploinsufficiency causes chronic obstructive pulmonary disease. Journal of Medical Genetics, 2011, 48, 705-709.	1.5	54
47	Autosomal recessive pure hair andÂnail ectodermal dysplasia linked toÂchromosome 12p11.1-q14.3 without KRTHB5 gene mutation. European Journal of Dermatology, 2010, 20, 443-446.	0.3	10
48	A Single-Nucleotide Deletion in the POMP 5â€² UTR Causes a Transcriptional Switch and Altered Epidermal Proteasome Distribution in KLICK Genodermatosis. American Journal of Human Genetics, 2010, 86, 596-603.	2.6	79
49	A Single-Nucleotide Deletion in the POMP 5â€² UTR Causes a Transcriptional Switch and Altered Epidermal Proteasome Distribution in KLICK Genodermatosis. American Journal of Human Genetics, 2010, 86, 655.	2.6	0
50	Cooperative effect of ribosomal protein s19 and Pim-1 kinase on murine c-Myc expression and myeloid/erythroid cellularity. Journal of Molecular Medicine, 2010, 88, 39-46.	1.7	8
51	Vascular endothelial growth factor B controls endothelial fatty acid uptake. Nature, 2010, 464, 917-921.	13.7	423
52	Familiar Meniere's disease restricted to 1.48â€‰Mb on chromosome 12p12.3 by allelic and haplotype association. Journal of Human Genetics, 2010, 55, 834-837.	1.1	26
53	Cooperative effect of ribosomal protein s19 and Pim-1 kinase on murine c-Myc expression and myeloid/erythroid cellularity. Nature Precedings, 2009, , .	0.1	0
54	Vascular Endothelial Growth Factor-B Induces Myocardium-Specific Angiogenesis and Arteriogenesis via Vascular Endothelial Growth Factor Receptor-1â€“ and Neuropilin Receptor-1â€“Dependent Mechanisms. Circulation, 2009, 119, 845-856.	1.6	172

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55	A chromosome 10 variant with a 12 Mb inversion [inv(10)(q11.22q21.1)] identical by descent and frequent in the Swedish population. American Journal of Medical Genetics, Part A, 2009, 149A, 380-386.	0.7	13
56	WNT10A missense mutation associated with a complete Odonto-Onycho-Dermal Dysplasia syndrome. European Journal of Human Genetics, 2009, 17, 1600-1605.	1.4	64
57	Mutations in the Fatty Acid Transport Protein 4 Gene Cause the Ichthyosis Prematurity Syndrome. American Journal of Human Genetics, 2009, 85, 248-253.	2.6	147
58	Alpha-cardiac actin mutations produce atrial septal defects. Human Molecular Genetics, 2008, 17, 256-265.	1.4	128
59	Combined Disruptions of the Ribosomal Protein s19 and Pim1 Kinase Genes Are Associated with Increased Myeloid/Erythroid Cellularity and Reduced Apoptosis. Blood, 2008, 112, 3097-3097.	0.6	0
60	Congenital ichthyosis: mutations in ichthyin are associated with specific structural abnormalities in the granular layer of epidermis. Journal of Medical Genetics, 2007, 44, 615-620.	1.5	59
61	Variant Phenotype of Best Vitelliform Macular Dystrophy Associated with Compound Heterozygous Mutations inVMD2. Ophthalmic Genetics, 2006, 27, 51-56.	0.5	74
62	Familial MÃ©niÃ¨re's Disease in Five Generations. Otology and Neurotology, 2006, 27, 681-686.	0.7	42
63	A founder mutation for ichthyosis prematurity syndrome restricted to 76Âkb by haplotype association. Journal of Human Genetics, 2006, 51, 864-871.	1.1	18
64	A Meniere's disease gene linked to chromosome 12p12.3. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 463-467.	1.1	56
65	Mutations in the gene encoding fibroblast growth factor 10 are associated with aplasia of lacrimal and salivary glands. Nature Genetics, 2005, 37, 125-128.	9.4	144
66	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. European Journal of Human Genetics, 2005, 13, 928-934.	1.4	14
67	Assignment of the locus for ichthyosis prematurity syndrome to chromosome 9q33.3-34.13. Journal of Medical Genetics, 2004, 41, 208-212.	1.5	26
68	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. British Journal of Haematology, 2002, 119, 261-264.	1.2	12
69	The gene encoding ribosomal protein S19 is mutated in Diamond-Blackfan anaemia. Nature Genetics, 1999, 21, 169-175.	9.4	747
70	Truncating ribosomal protein S19 mutations and variable clinical expression in Diamond-Blackfan anemia. Human Genetics, 1999, 105, 496-500.	1.8	20
71	Truncating ribosomal protein S19 mutations and variable clinical expression in Diamond-Blackfan anemia. Human Genetics, 1999, 105, 496-500.	1.8	40