Niklas Dahl

List of Publications by Year
in descending order

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120 papers	6,377 citations	126907 33 h-index	69250 77 g-index
130 all docs	130 docs citations	130 times ranked	8607 citing authors

#	Article	IF	Citations
1	A gene encoding a liver-specific ABC transporter is mutated in progressive familial intrahepatic cholestasis. Nature Genetics, 1998, 20, 233-238.	21.4	968
2	The gene encoding ribosomal protein S19 is mutated in Diamond-Blackfan anaemia. Nature Genetics, 1999, 21, 169-175.	21.4	747
3	A gene mutated in Xâ€“linked myotubular myopathy defines a new putative tyrosine phosphatase family conserved in yeast. Nature Genetics, 1996, 13, 175-182.	21.4	586
4	Diagnosing and treating Diamond Blackfan anaemia: results of an international clinical consensus conference. British Journal of Haematology, 2008, 142, 859-876.	2.5	408
5	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. Annals of Neurology, 1999, 45, 200-206.	5.3	371
6	The ribosomal basis of diamond-blackfan anemia: mutation and database update. Human Mutation, 2010, 31, 1269-1279.	2.5	202
7	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	21.4	196
8	Mutations in the Fatty Acid Transport Protein 4 Gene Cause the Ichthyosis Prematurity Syndrome. American Journal of Human Genetics, 2009, 85, 248-253.	6.2	147
9	A Missense Mutation in the Aggrecan C-type Lectin Domain Disrupts Extracellular Matrix Interactions and Causes Dominant Familial Osteochondritis Dissecans. American Journal of Human Genetics, 2010, 86, 126-137.	6.2	140
10	Isolated oligodontia associated with mutations in EDARADD, AXIN2, MSX1, and PAX9 genes. American Journal of Medical Genetics, Part A, 2011, 155, 1616-1622.	1.2	124
11	Diamond-Blackfan anaemia: genetic homogeneity for a gene on chromosome 19q13 restricted to 1.8 Mb. Nature Genetics, 1997, 16, 368-371.	21.4	93
12	Welander Distal Myopathy Caused by an Ancient Founder Mutation inTIA1Associated with Perturbed Splicing. Human Mutation, 2013, 34, n/a-n/a.	2.5	91
13	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. Cell Reports, 2018, 24, 3441-3454.e12.	6.4	91
14	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.	6.2	79
15	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.	1.2	69
16	WNT10A missense mutation associated with a complete Odonto-Onycho-Dermal Dysplasia syndrome. European Journal of Human Genetics, 2009, 17, 1600-1605.	2.8	64
17	A combined approach for single-cell mRNA and intracellular protein expression analysis. Communications Biology, 2021, 4, 624.	4.4	64
18	Abolished InsP3R2 function inhibits sweat secretion in both humans and mice. Journal of Clinical Investigation, 2014, 124, 4773-4780.	8.2	63

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19	Detailed analysis of HTT repeat elements in human blood using targeted amplification-free long-read sequencing. Human Mutation, 2018, 39, 1262-1272.	2.5	62
20	Mutations in Frizzled 6 Cause Isolated Autosomal-Recessive Nail Dysplasia. American Journal of Human Genetics, 2011, 88, 852-860.	6.2	58
21	Disheveled regulates precoupling of heterotrimeric G proteins to Frizzled 6. FASEB Journal, 2014, 28, 2293-2305.	0.5	58
22	ACTG2variants impair actin polymerization in sporadic Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. Human Molecular Genetics, 2016, 25, 571-583.	2.9	56
23	Fibroblast growth factor 10 haploinsufficiency causes chronic obstructive pulmonary disease. Journal of Medical Genetics, 2011, 48, 705-709.	3.2	54
24	Altered paracellular cation permeability due to a rare CLDN10B variant causes anhidrosis and kidney damage. PLoS Genetics, 2017, 13, e1006897.	3.5	50
25	Untangling the Phenotypic Heterogeneity of Diamond Blackfan Anemia. Seminars in Hematology, 2011, 48, 124-135.	3.4	48
26	SPG11 mutations cause Kjellin syndrome, a hereditary spastic paraplegia with thin corpus callosum and central retinal degeneration. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 984-992.	1.7	46
27	Genomic duplications mediate overexpression of lamin B1 in adult-onset autosomal dominant leukodystrophy (ADLD) with autonomic symptoms. Neurogenetics, 2011, 12, 65-72.	1.4	43
28	SNX10 gene mutation leading to osteopetrosis with dysfunctional osteoclasts. Scientific Reports, 2017, 7, 3012.	3.3	43
29	Down-regulation of progesterone receptor membrane component 1 (PGRMC1) in peripheral nucleated blood cells associated with premature ovarian failure (POF) and polycystic ovary syndrome (PCOS). Reproductive Biology and Endocrinology, 2010, 8, 58.	3.3	42
30	Ribosomal protein S19 and S24 insufficiency cause distinct cell cycle defects in Diamondâ€“Blackfan anemia. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1036-1042.	3.8	41
31	MuSK: a new target for lethal fetal akinesia deformation sequence (FADS). Journal of Medical Genetics, 2015, 52, 195-202.	3.2	41
32	Recurrent <scp>GATA</scp>1 mutations in <scp>D</scp>iamondâ€<scp>B</scp>lackfan anaemia. British Journal of Haematology, 2014, 166, 949-951.	2.5	40
33	Single cell analysis of autism patient with bi-allelic NRXN1-alpha deletion reveals skewed fate choice in neural progenitors and impaired neuronal functionality. Experimental Cell Research, 2019, 383, 111469.	2.6	39
34	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.	4.0	36
35	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.	2.8	35
36	L<scp>MNB</scp>1â€related autosomalâ€dominant leukodystrophy: Clinical and radiological course. Annals of Neurology, 2015, 78, 412-425.	5.3	35

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37	Analysis of <scp> LMNB </scp> 1 Duplications in Autosomal Dominant Leukodystrophy Provides Insights into Duplication Mechanisms and Alleleâ€Specific Expression. Human Mutation, 2013, 34, 1160-1171.	2.5	33
38	Transcriptomes of Dravet syndrome iPSC derived GABAergic cells reveal dysregulated pathways for chromatin remodeling and neurodevelopment. Neurobiology of Disease, 2019, 132, 104583.	4.4	32
39	Frizzled6 Deficiency Disrupts the Differentiation Process of Nail Development. Journal of Investigative Dermatology, 2013, 133, 1990-1997.	0.7	30
40	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29
41	siRNA Silencing of Proteasome Maturation Protein (POMP) Activates the Unfolded Protein Response and Constitutes a Model for KLICK Genodermatosis. PLoS ONE, 2012, 7, e29471.	2.5	29
42	A novel missense mutation in the EDA gene associated with X-linked recessive isolated hypodontia. Journal of Human Genetics, 2008, 53, 894-898.	2.3	28
43	Genome-wide sequencing for the identification of rearrangements associated with Tourette syndrome and obsessive-compulsive disorder. BMC Medical Genetics, 2012, 13, 123.	2.1	28
44	FATP4 missense and nonsense mutations cause similar features in Ichthyosis Prematurity Syndrome. BMC Research Notes, 2011, 4, 90.	1.4	27
45	Phenotypic expansion of visceral myopathy associated with ACTG2 tandem base substitution. European Journal of Human Genetics, 2015, 23, 1679-1683.	2.8	27
46	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.	2.8	27
47	NÎ±-Tosyl-l-phenylalanine Chloromethyl Ketone Induces Caspase-dependent Apoptosis in Transformed Human B Cell Lines with Transcriptional Down-regulation of Anti-apoptotic HS1-associated Protein X-1. Journal of Biological Chemistry, 2009, 284, 27827-27837.	3.4	26
48	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.	2.3	26
49	Clinical utility gene card for: Diamond â€“ Blackfan Anemia â€“ update 2013. European Journal of Human Genetics, 2013, 21, 1187-1187.	2.8	26
50	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.	1.3	22
51	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
52	Transcriptome Profiling Reveals Degree of Variability in Induced Pluripotent Stem Cell Lines: Impact for Human Disease Modeling. Cellular Reprogramming, 2015, 17, 327-337.	0.9	21
53	Generation of human iPS cell line CTL07-II from human fibroblasts, under defined and xeno-free conditions. Stem Cell Research, 2016, 17, 474-478.	0.7	21
54	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

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55	A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324.	2.0	21
56	Posttranscriptional downâ€regulation of small ribosomal subunit proteins correlates with reduction of 18S rRNA in RPS19 deficiency. FEBS Letters, 2009, 583, 2049-2053.	2.8	20
57	Methods of Reprogramming to Induced Pluripotent Stem Cell Associated with Chromosomal Integrity and Delineation of a Chromosome 5q Candidate Region for Growth Advantage. Stem Cells and Development, 2015, 24, 2032-2040.	2.1	20
58	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.	4.1	20
59	Variability in noise susceptibility in a Swedish population: the role of 35delG mutation in the connexin 26 (GJB2) gene. Audiological Medicine, 2004, 2, 123-130.	0.4	19
60	Somatic mosaicism for chromosome X and Y aneuploidies in monozygotic twins heterozygous for sickle cell disease mutation. American Journal of Medical Genetics, Part A, 2010, 152A, 2595-2598.	1.2	19
61	Novel SACS mutations associated with intellectual disability, epilepsy and widespread supratentorial abnormalities. Journal of the Neurological Sciences, 2016, 371, 105-111.	0.6	19
62	Multiple epiphyseal dysplasia. Monthly Notices of the Royal Astronomical Society: Letters, 2009, 80, 711-715.	3.3	18
63	A balanced reciprocal translocation t(5;7)(q14;q32) associated with autistic disorder: Molecular analysis of the chromosome 7 breakpoint. American Journal of Medical Genetics Part A, 2001, 105, 729-736.	2.4	17
64	Ichthyin/NIPAL4 localizes to keratins and desmosomes in epidermis and Ichthyin mutations affect epidermal lipid metabolism. Archives of Dermatological Research, 2012, 304, 377-386.	1.9	17
65	Abnormal primary and permanent dentitions with ectodermal symptoms predict WNT10A deficiency. BMC Medical Genetics, 2016, 17, 88.	2.1	17
66	Deletion of the hunter gene and both DXS466 and DXS304 in a patient with mucopolysaccharidosis type II. American Journal of Medical Genetics Part A, 1992, 44, 100-103.	2.4	16
67	Two novel mutations (L32P) and (G85N) among five different missense mutations in six Danish families with Fabry's disease. Human Mutation, 1995, 5, 277-278.	2.5	16
68	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
69	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.	2.5	16
70	Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 739-748.	6.2	15
71	Compound heterozygous HAX1 mutations in a Swedish patient with severe congenital neutropenia and no neurodevelopmental abnormalities. Pediatric Blood and Cancer, 2009, 53, 1143-1146.	1.5	14
72	Whole exome sequencing identifiesLRP1as a pathogenic gene in autosomal recessive keratosis pilaris atrophicans. Journal of Medical Genetics, 2015, 52, 599-606.	3.2	14

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73	5â€²UTR Variants of Ribosomal Protein S19 Transcript Determine Translational Efficiency: Implications for Diamond-Blackfan Anemia and Tissue Variability. PLoS ONE, 2011, 6, e17672.	2.5	14
74	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.	1.2	13
75	Stereocilin gene variants associated with episodic vertigo: expansion of the DFNB16 phenotype. European Journal of Human Genetics, 2018, 26, 1871-1874.	2.8	12
76	Bedside diagnosis of rippling muscle disease in CAV3 p.A46T mutation carriers. Muscle and Nerve, 2010, 41, 751-757.	2.2	11
77	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.	2.2	11
78	Rapid detection of a mutation hotâ€spot in the human androgen receptor. Clinical Genetics, 1996, 50, 202-205.	2.0	10
79	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.	0.6	10
80	Ribosomal protein S19 binds to its own mRNA with reduced affinity in Diamond-Blackfan anemia. Blood Cells, Molecules, and Diseases, 2010, 45, 23-28.	1.4	10
81	A novel mutation in the Lipase H gene underlies autosomal recessive hypotrichosis and woolly hair. Scientific Reports, 2012, 2, 730.	3.3	10
82	Aniridia with PAX6 mutations and narcolepsy. Journal of Sleep Research, 2020, 29, e12982.	3.2	10
83	Targeted Resequencing and Analysis of the Diamond-Blackfan Anemia Disease Locus RPS19. PLoS ONE, 2009, 4, e6172.	2.5	10
84	Inherited mosaicism for the supernumerary marker chromosome in cat eye syndrome: Interâ€and intraâ€individual variation and correlation to the phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 1111-1117.	1.2	9
85	Generation of three human induced pluripotent stem cell (iPSC) lines from three patients with Dravet syndrome carrying distinct SCN1A gene mutations. Stem Cell Research, 2019, 39, 101523.	0.7	9
86	GATA-1 Defects in Diamondâ€“Blackfan Anemia: Phenotypic Characterization Points to a Specific Subset of Disease. Genes, 2022, 13, 447.	2.4	9
87	PRENATAL DIAGNOSIS OF X-LINKED MYOTUBULAR MYOPATHY: STRATEGIES USING NEW AND TIGHTLY LINKED DNA MARKERS. , 1996, 16, 231-237.		8
88	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.	3.9	8
89	Whole exome sequencing identifies novel variant underlying hereditary spastic paraplegia in consanguineous Pakistani families. Journal of Clinical Neuroscience, 2019, 67, 19-23.	1.5	8
90	Study of individuals possibly affected with the fragile X syndrome in a large swedish family in the 18th to 20th centuries. American Journal of Medical Genetics Part A, 1992, 43, 353-354.	2.4	7

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91	Infantile autismâ€"fragile X: Molecular findings support genetic heterogeneity. American Journal of Medical Genetics Part A, 1992, 44, 830-833.	2.4	7
92	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Frontiers in Genetics, 2019, 10, 896.	2.3	7
93	A novel mutation (C201R) in the transmembrane domain of connexin 32 in severe x-linked charcot-marie-tooth disease. Human Mutation, 1998, 11, S8-S9.	2.5	6
94	New perspectives on the dynamic behaviour of oral lichen planus. European Journal of Dermatology, 2012, 22, 178-181.	0.6	6
95	Mowat-Wilson syndrome: Generation of two human iPS cell lines (UUIGPi004A and UUIGPi005A) from siblings with a truncating ZEB2 gene variant. Stem Cell Research, 2019, 39, 101518.	0.7	5
96	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.	1.6	5
97	Phenotypic variability in chorea-acanthocytosis associated with novel VPS13A mutations. Neurology: Genetics, 2020, 6, e426.	1.9	5
98	Clinical utility gene card for: Diamond Blackfan anemia. European Journal of Human Genetics, 2011, 19, 615-615.	2.8	4
99	Generation of a human Neurochondrin deficient iPSC line KICRi002-A-3 using CRISPR/Cas9. Stem Cell Research, 2020, 44, 101758.	0.7	4
100	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.	1.9	4
101	Molecular characterization of a DNA probe, U6.2, located close to the fragile X locus. American Journal of Medical Genetics Part A, 1991, 38, 380-383.	2.4	3
102	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.	0.7	3
103	Linkage analysis of the fragile X syndrome using a new DNA marker U6.2 defining locus DXS304. American Journal of Medical Genetics Part A, 1991, 38, 322-327.	2.4	2
104	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.	0.6	2
105	Primary microcephaly, primordial dwarfism, and brachydactyly in adult cases with biallelic skipping of RTTN exon 42. Human Mutation, 2019, 40, 899-903.	2.5	2
106	Whole genome sequencing of familial isolated oesophagus atresia uncover shared structural variants. BMC Medical Genomics, 2020, 13, 85.	1.5	2
107	Friedreich's ataxia: Point mutations and clinical presentation of compound heterozygotes. , 1999, 45, 200.		2
108	Syndromic RNA polymerase II insufficiency: Generation of a human induced pluripotent stem cell line (UUIGPi002A-5) with a heterozygous disruption of POLR2A. Stem Cell Research, 2021, 57, 102577.	0.7	2

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109	Generation of two human iPSC lines (UUIGPi013-A and UUIPGi014-A) from cases with Down syndrome and full trisomy for chromosome 21 (T21). Stem Cell Research, 2020, 49, 102081.	0.7	1
110	Generation of human induced pluripotent stem cell (iPSC) lines (UUMCBi001-A, UUMCBi002-A) from two healthy donors. Stem Cell Research, 2021, 50, 102114.	0.7	1
111	Identification of a novel variant in GPR56/ADGRG1 gene through whole exome sequencing in a consanguineous Pakistani family. Journal of Clinical Neuroscience, 2021, 94, 8-12.	1.5	1
112	Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. Frontiers in Genetics, 2021, 12, 803683.	2.3	1
113	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
114	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.	6.2	0
115	Incontinentia pigmenti: Generation of an IKBKG deficient human iPSC line (KICRi002-A-1) on a 46,XY background using CRISPR/Cas9. Stem Cell Research, 2020, 44, 101739.	0.7	0
116	Hematopoietic Mechanism in Diamond-Blackfan Anemia: Late Erythroid Development Is Not Affected by Ribosomal Protein S19 Deficiency.. Blood, 2004, 104, 719-719.	1.4	0
117	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.	1.4	0
118	Ichthyosis Prematurity Syndrome. , 2016, , 1445-1447.		0
119	De Novo Sequence and Copy Number Variants are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. SSRN Electronic Journal, 0, , .	0.4	0
120	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.	0.7	0