Christian P Schaaf

List of Publications by Year
in descending order

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123 papers	6,752 citations	66234 42 h-index	71532 76 g-index
135 all docs	135 docs citations	135 times ranked	11397 citing authors

#	Article	IF	Citations
1	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
2	SHANK3 overexpression causes manic-like behaviour with unique pharmacogenetic properties. Nature, 2013, 503, 72-77.	13.7	323
3	Autism and other neuropsychiatric symptoms are prevalent in individuals with MeCP2 duplication syndrome. Annals of Neurology, 2009, 66, 771-782.	2.8	271
4	Truncating mutations of MAGEL2 cause Prader-Willi phenotypes and autism. Nature Genetics, 2013, 45, 1405-1408.	9.4	258
5	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	1.1	225
6	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
7	Quantitative real-time imaging of glutathione. Nature Communications, 2017, 8, 16087.	5.8	192
8	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	1.1	191
9	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
10	USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder. Molecular Cell, 2015, 59, 956-969.	4.5	175
11	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. Nature Genetics, 2009, 41, 1269-1271.	9.4	171
12	Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. Human Molecular Genetics, 2011, 20, 3366-3375.	1.4	149
13	Copy Number and SNP Arrays in Clinical Diagnostics. Annual Review of Genomics and Human Genetics, 2011, 12, 25-51.	2.5	143
14	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	1.1	143
15	Solving the Autism Puzzle a Few Pieces at a Time. Neuron, 2011, 70, 806-808.	3.8	134
16	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
17	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
18	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	2.4	120

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19	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	9.4	113
20	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	1.1	111
21	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. European Journal of Human Genetics, 2011, 19, 102-107.	1.4	104
22	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	13.5	103
23	Phenotypic spectrum and genotypeâ€“phenotype correlations of NRXN1 exon deletions. European Journal of Human Genetics, 2012, 20, 1240-1247.	1.4	99
24	The human clinical phenotypes of altered CHRNA7 copy number. Biochemical Pharmacology, 2015, 97, 352-362.	2.0	97
25	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	1.1	94
26	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	2.6	92
27	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	7.7	83
28	Nicotinic acetylcholine receptors in human genetic disease. Genetics in Medicine, 2014, 16, 649-656.	1.1	82
29	NUDT21-spanning CNVs lead to neuropsychiatric disease and altered MeCP2 abundance via alternative polyadenylation. ELife, 2015, 4, .	2.8	74
30	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	9.4	73
31	Early-onset severe neuromuscular phenotype associated with compound heterozygosity for OPA1 mutations. Molecular Genetics and Metabolism, 2011, 103, 383-387.	0.5	71
32	Prader-Willi Syndrome and Schaaf-Yang Syndrome: Neurodevelopmental Diseases Intersecting at the MAGEL2 Gene. Diseases (Basel, Switzerland), 2016, 4, 2.	1.0	69
33	The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. Journal of Autism and Developmental Disorders, 2017, 47, 549-562.	1.7	68
34	Schaafâ€¥ang syndrome overview: Report of 78 individuals. American Journal of Medical Genetics, Part A, 2018, 176, 2564-2574.	0.7	66
35	Otud7a Knockout Mice Recapitulate Many Neurological Features of 15q13.3 Microdeletion Syndrome. American Journal of Human Genetics, 2018, 102, 296-308.	2.6	65
36	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	1.1	64

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37	Identification of incestuous parental relationships by SNP-based DNA microarrays. Lancet, The, 2011, 377, 555-556.	6.3	57
38	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	2.6	56
39	Desmosterolosisâ€"phenotypic and molecular characterization of a third case and review of the literature. American Journal of Medical Genetics, Part A, 2011, 155, 1597-1604.	0.7	52
40	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52
41	Functional Consequences of CHRNA7 Copy-Number Alterations in Induced Pluripotent Stem Cells and Neural Progenitor Cells. American Journal of Human Genetics, 2017, 101, 874-887.	2.6	50
42	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	3.6	50
43	The Genetics of Autism Spectrum Disorders â€“ A Guide for Clinicians. Current Psychiatry Reports, 2013, 15, 334.	2.1	49
44	Autism genetics â€“ an overview. Prenatal Diagnosis, 2017, 37, 14-30.	1.1	49
45	An estimation of the prevalence of genomic disorders using chromosomal microarray data. Journal of Human Genetics, 2018, 63, 795-801.	1.1	49
46	Expanding the clinical spectrum of the 16p11.2 chromosomal rearrangements: three patients with syringomyelia. European Journal of Human Genetics, 2011, 19, 152-156.	1.4	47
47	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	2.6	45
48	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. Genetics in Medicine, 2016, 18, 1111-1118.	1.1	45
49	Chrna7 deficient mice manifest no consistent neuropsychiatric and behavioral phenotypes. Scientific Reports, 2017, 7, 39941.	1.6	43
50	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	1.1	41
51	Mutations in ASH1L confer susceptibility to Tourette syndrome. Molecular Psychiatry, 2020, 25, 476-490.	4.1	41
52	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	1.1	40
53	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617.	9.4	40
54	Magel2 knockout mice manifest altered social phenotypes and a deficit in preference for social novelty. Genes, Brain and Behavior, 2017, 16, 592-600.	1.1	39

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55	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. European Journal of Human Genetics, 2014, 22, 1071-1076.	1.4	37
56	Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review. BMC Medical Genetics, 2015, 16, 12.	2.1	37
57	Novel mutation of the WDR45 gene causing betaâ€propeller proteinâ€associated neurodegeneration. Movement Disorders, 2014, 29, 574-575.	2.2	34
58	Clinical and molecular characterization of de novo loss of function variants in HNRNPU. American Journal of Medical Genetics, Part A, 2017, 173, 2680-2689.	0.7	34
59	Human subtelomeric copy number gains suggest a DNA replication mechanism for formation: beyond breakageâ€“fusionâ€“bridge for telomere stabilization. Human Genetics, 2012, 131, 1895-1910.	1.8	32
60	Hormonal, metabolic and skeletal phenotype of Schaaf-Yang syndrome: a comparison to Prader-Willi syndrome. Journal of Medical Genetics, 2018, 55, 307-315.	1.5	32
61	BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. American Journal of Human Genetics, 2020, 107, 1096-1112.	2.6	32
62	Combination of whole exome sequencing and animal modeling identifies TMPRSS9 as a candidate gene for autism spectrum disorder. Human Molecular Genetics, 2020, 29, 459-470.	1.4	32
63	Improvement of regressive autism symptoms in a child with TMLHE deficiency following carnitine supplementation. American Journal of Medical Genetics, Part A, 2015, 167, 2162-2167.	0.7	30
64	Genetic causes of optic nerve hypoplasia. Journal of Medical Genetics, 2017, 54, 441-449.	1.5	30
65	Mitochondrial involvement in a Bosch-Boonstra-Schaaf optic atrophy syndrome patient with a novel de novo NR2F1 gene mutation. Journal of Human Genetics, 2018, 63, 525-528.	1.1	27
66	Phenotypic expansion of <scp>Boschâ€“Boonstraâ€“Schaaf</scp> optic atrophy syndrome and further evidence for genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2020, 182, 1426-1437.	0.7	27
67	Facebook Support Groups for Rare Pediatric Diseases: Quantitative Analysis. JMIR Pediatrics and Parenting, 2020, 3, e21694.	0.8	26
68	Novel interaction partners of the TPR/MET tyrosine kinase. FASEB Journal, 2005, 19, 1-21.	0.2	25
69	mTOR and autophagy pathways are dysregulated in murine and human models of Schaaf-Yang syndrome. Scientific Reports, 2019, 9, 15935.	1.6	24
70	Diagnosis and genetics of alacrima. Clinical Genetics, 2018, 94, 54-60.	1.0	17
71	MAGEL2 and Oxytocinâ€”Implications in Prader-Willi Syndrome and Beyond. Biological Psychiatry, 2015, 78, 78-80.	0.7	16
72	Magel2 Modulates Bone Remodeling and Mass in Prader-Willi Syndrome by Affecting Oleoyl Serine Levels and Activity. Journal of Bone and Mineral Research, 2019, 34, 93-105.	3.1	16

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73	Next Generation Sequencing of 134 Children with Autism Spectrum Disorder and Regression. Genes, 2020, 11, 853.	1.0	16
74	Phenotypic and Imaging Spectrum Associated With WDR45. Pediatric Neurology, 2020, 109, 56-62.	1.0	16
75	Truncating variants in UBAP1 associated with childhoodâ€onset nonsyndromic hereditary spastic paraplegia. Human Mutation, 2020, 41, 632-640.	1.1	15
76	Assigning evidence to actionability: An introduction to variant interpretation in precision cancer medicine. Genes Chromosomes and Cancer, 2022, 61, 303-313.	1.5	15
77	The adult phenotype of Schaaf-Yang syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 294.	1.2	14
78	Mecp2Deletion from Cholinergic Neurons Selectively Impairs Recognition Memory and Disrupts Cholinergic Modulation of the Perirhinal Cortex. ENeuro, 2019, 6, ENEURO.0134-19.2019.	0.9	14
79	A partial MECP2 duplication in a mildly affected adult male: a putative role for the 3' untranslated region in the MECP2 duplication phenotype. BMC Medical Genetics, 2012, 13, 71.	2.1	12
80	CHRNA7 copy number gains are enriched in adolescents with major depressive and anxiety disorders. Journal of Affective Disorders, 2018, 239, 247-252.	2.0	12
81	Coâ€occurring defect analysis: A platform for analyzing birth defect coâ€occurrence in registries. Birth Defects Research, 2019, 111, 1356-1364.	0.8	12
82	Side Effects and Behavioral Outcomes Following High-Dose Carnitine Supplementation Among Young Males With Autism Spectrum Disorder: A Pilot Study. Global Pediatric Health, 2019, 6, 2333794X1983069.	0.3	12
83	Nr2f1 heterozygous knockout mice recapitulate neurological phenotypes of Bosch-Boonstra-Schaaf optic atrophy syndrome and show impaired hippocampal synaptic plasticity. Human Molecular Genetics, 2020, 29, 705-715.	1.4	12
84	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	1.1	12
85	Molecular characterization of HDAC8 deletions in individuals with atypical Cornelia de Lange syndrome. Journal of Human Genetics, 2018, 63, 349-356.	1.1	10
86	Neurocognitive and Neurobehavioral Phenotype of Youth with Schaaf-Yang Syndrome. Journal of Autism and Developmental Disorders, 2020, 50, 2491-2500.	1.7	10
87	The expanding clinical phenotype of germline ABL1 â€associated congenital heart defects and skeletal malformations syndrome. Human Mutation, 2020, 41, 1738-1744.	1.1	10
88	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	1.1	10
89	Pathophysiological Heterogeneity of the BBSOA Neurodevelopmental Syndrome. Cells, 2022, 11, 1260.	1.8	10
90	CHRNA7 Deletions are Enriched in Risperidone-Treated Children and Adolescents. Journal of Child and Adolescent Psychopharmacology, 2017, 27, 908-915.	0.7	9

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91	Chronic intestinal pseudo-obstruction syndrome and gastrointestinal malrotation in an infantwith schaaf-yang syndrome - Expanding the phenotypic spectrum. European Journal of Medical Genetics, 2018, 61, 627-630.	0.7	9
92	Birth defects that coâ€•occur with nonâ€•syndromic gastroschisis and omphalocele. American Journal of Medical Genetics, Part A, 2020, 182, 2581-2593.	0.7	9
93	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
94	Intragenic deletions of the IGF1 receptor gene in five individuals with psychiatric phenotypes and developmental delay. European Journal of Human Genetics, 2013, 21, 1304-1307.	1.4	8
95	Birth defect co-occurrence patterns in the Texas Birth Defects Registry. Pediatric Research, 2022, 91, 1278-1285.	1.1	8
96	The importance of phase analysis in multiexon copy number variation detected by aCGH in autosomal recessive disorder loci. , 2017, 173, 2485-2488.		7
97	Polysomnographic characteristics and sleepâ€disordered breathing in Schaafâ€Yang syndrome. Pediatric Pulmonology, 2020, 55, 3162-3167.	1.0	7
98	Germline testing for homologous recombination repair genesâ€”opportunities and challenges. Genes Chromosomes and Cancer, 2021, 60, 332-343.	1.5	7
99	Parental perceptions of genetic testing for children with autism spectrum disorders. American Journal of Medical Genetics, Part A, 2022, 188, 178-186.	0.7	7
100	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	1.5	7
101	Facebook Support Groups for Pediatric Rare Diseases: Cross-Sectional Study to Investigate Opportunities, Limitations, and Privacy Concerns. JMIR Pediatrics and Parenting, 2022, 5, e31411.	0.8	7
102	Imprinting: the Achilles heel of trio-based exome sequencing. Genetics in Medicine, 2016, 18, 1163-1164.	1.1	6
103	Which genes to assess in the NGS diagnostics of intellectual disability? The case for a consensus database-driven and expert-curated approach. Molecular and Cellular Probes, 2019, 45, 84-88.	0.9	6
104	Genomic newborn screening: Proposal of a <scp>twoâ€stage</scp> approach. Journal of Inherited Metabolic Disease, 2021, 44, 518-520.	1.7	6
105	A retrospective analysis of growth hormone therapy in children with <scp>Schaafâ€“Yang</scp> syndrome. Clinical Genetics, 2021, 100, 298-307.	1.0	5
106	MiRNA-149 as a Candidate for Facial Clefting and Neural Crest Cell Migration. Journal of Dental Research, 2022, 101, 323-330.	2.5	5
107	Hans Gunther and his disease. Photodermatology Photoimmunology and Photomedicine, 2007, 23, 261-263.	0.7	4
108	Patterns of co-occurring birth defects among infants with hypospadias. Journal of Pediatric Urology, 2021, 17, 64.e1-64.e8.	0.6	4

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109	Oâ€™Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum. Journal of Medical Genetics, 2021, , jmedgenet-2020-107470.	1.5	4
110	A Comprehensive Assessment of Co-occurring Birth Defects among Infants with Non-Syndromic Anophthalmia or Microphthalmia. Ophthalmic Epidemiology, 2021, 28, 428-435.	0.8	4
111	Assessment of Cognitive Outcome Measures in Teenagers with 15q13.3 Microdeletion Syndrome. Journal of Autism and Developmental Disorders, 2016, 46, 1455-1463.	1.7	3
112	Next Generation Sequencing in Autism Spectrum Disorder. , 2018, 2, 1-1.		3
113	Genetic counseling and the role of genetic counselors in the United States. Medizinische Genetik, 2021, 33, 29-34.	0.1	3
114	Identification of complex chromosome 18 rearrangements by FISH and array CGH in two patients with apparent isochromosome 18q. American Journal of Medical Genetics, Part A, 2011, 155, 1465-1468.	0.7	2
115	Patterns of congenital anomalies among individuals with trisomy 13 in Texas. American Journal of Medical Genetics, Part A, 2021, 185, 1787-1793.	0.7	2
116	Translational pediatrics: clinical perspective for Phelanâ€“McDermid syndrome and autism research. Pediatric Research, 2022, 92, 373-377.	1.1	2
117	From newborn screening to genomic medicine: challenges and suggestions on how to incorporate genomic newborn screening in public health programs. Medizinische Genetik, 2022, 34, 13-20.	0.1	2
118	Spontaneous hyperactivity in Ash1l mutant mice, a new model for Tourette syndrome. Molecular Psychiatry, 2020, 25, 241-242.	4.1	1
119	Response to Briuglia et al.. Genetics in Medicine, 2021, 23, 423-424.	1.1	1
120	Brain Network Analysis of EEG Recordings Can Be Used to Assess Cognitive Function in Teenagers With 15q13.3 Microdeletion Syndrome. Frontiers in Neuroscience, 2021, 15, 622329.	1.4	1
121	Genetic Variation in LRP1 Associates with Stanford Type B Aortic Dissection Risk and Clinical Outcome. Journal of Cardiovascular Development and Disease, 2022, 9, 14.	0.8	1
122	Phenotypic characterization of seven individuals with <scp>Marbachâ€“Schaaf</scp> neurodevelopmental syndrome. American Journal of Medical Genetics, Part A, 0, , .	0.7	1
123	15q13.3 Microdeletion Syndrome. , 2021, , 1-3.		0