Carol J Saunders

List of Publications by Year
in descending order

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75 papers	3,519 citations	147801 31 h-index	149698 56 g-index
78 all docs	78 docs citations	78 times ranked	7145 citing authors

#	Article	IF	Citations
1	Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. Science Translational Medicine, 2014, 6, 265ra168.	12.4	440
2	Early-onset lymphoproliferation and autoimmunity caused by germline STAT3 gain-of-function mutations. Blood, 2015, 125, 591-599.	1.4	436
3	Whole-genome sequencing for identification of Mendelian disorders in critically ill infants: a retrospective analysis of diagnostic and clinical findings. Lancet Respiratory Medicine,the, 2015, 3, 377-387.	10.7	322
4	A 26-hour system of highly sensitive whole genome sequencing for emergency management of genetic diseases. Genome Medicine, 2015, 7, 100.	8.2	237
5	The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. Npj Genomic Medicine, 2018, 3, 6.	3.8	156
6	Deep Sequencing of Patient Genomes for Disease Diagnosis: When Will It Become Routine?. Science Translational Medicine, 2011, 03, 87ps23.	12.4	98
7	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. Frontiers in Immunology, 2016, 7, 466.	4.8	80
8	A novel epileptic encephalopathy mutation in KCNB1 disrupts Kv2.1 ion selectivity, expression, and localization. Journal of General Physiology, 2015, 146, 399-410.	1.9	79
9	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	9.0	79
10	Exome sequencing reveals a pallidin mutation in a Hermansky-Pudlakâ€“like primary immunodeficiency syndrome. Blood, 2012, 119, 3185-3187.	1.4	76
11	Diagnosis of mitochondrial disorders by concomitant next-generation sequencing of the exome and mitochondrial genome. Genomics, 2013, 102, 148-156.	2.9	68
12	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	6.2	66
13	MMP21 is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates. Nature Genetics, 2015, 47, 1260-1263.	21.4	65
14	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	2.0	63
15	CLPB Variants Associated with Autosomal-Recessive Mitochondrial Disorder with Cataract, Neutropenia, Epilepsy, and Methylglutaconic Aciduria. American Journal of Human Genetics, 2015, 96, 258-265.	6.2	58
16	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
17	Spectrum of K_V2.1 Dysfunction in KCNB1â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2019, 86, 899-912.	5.3	52
18	Next-generation community genetics for low- and middle-income countries. Genome Medicine, 2012, 4, 25.	8.2	51

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19	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
20	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
21	Adopting orphans: comprehensive genetic testing of Mendelian diseases of childhood by next-generation sequencing. Expert Review of Molecular Diagnostics, 2011, 11, 855-868.	3.1	45
22	Novel exon 1 mutations in MECP2 implicate isoform MeCP2_e1 in classical Rett syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1019-1023.	1.2	44
23	De novoframeshift mutation in ASXL3 in a patient with global developmental delay, microcephaly, and craniofacial anomalies. BMC Medical Genomics, 2013, 6, 32.	1.5	43
24	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	6.2	42
25	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (TRIT1) gene. Human Mutation, 2017, 38, 511-516.	2.5	39
26	GPR37L1 modulates seizure susceptibility: Evidence from mouse studies and analyses of a human GPR37L1 variant. Neurobiology of Disease, 2017, 106, 181-190.	4.4	38
27	PCDH19â€related epileptic encephalopathy in a male mosaic for a truncating variant. American Journal of Medical Genetics, Part A, 2016, 170, 1585-1589.	1.2	37
28	Neonatal progeriod syndrome associated with biallelic truncating variants in POLR3A. American Journal of Medical Genetics, Part A, 2016, 170, 3343-3346.	1.2	37
29	Genomic answers for children: Dynamic analyses of >1000 pediatric rare disease genomes. Genetics in Medicine, 2022, 24, 1336-1348.	2.4	37
30	Loss of Function Variants in Human PNPLA8 Encoding Calcium-Independent Phospholipase A₂ Î³ Recapitulate the Mitochondriopathy of the Homologous Null Mouse. Human Mutation, 2015, 36, 301-306.	2.5	36
31	Clinical genome sequencing in an unbiased pediatric cohort. Genetics in Medicine, 2019, 21, 303-310.	2.4	36
32	Molecular diagnosis of infantile onset inflammatory bowel disease by exome sequencing. Genomics, 2013, 102, 442-447.	2.9	35
33	Comprehensive ZEB2 gene analysis for Mowatâ€“Wilson syndrome in a North American cohort: A suggested approach to molecular diagnostics. American Journal of Medical Genetics, Part A, 2009, 149A, 2527-2531.	1.2	33
34	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	8.5	32
35	Clinical detection of deletion structural variants in whole-genome sequences. Npj Genomic Medicine, 2016, 1, 16026.	3.8	29
36	A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. Journal of Inherited Metabolic Disease, 2017, 40, 853-860.	3.6	27

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37	A patient with polymerase E1 deficiency (POLE1): clinical features and overlap with DNA breakage/instability syndromes. BMC Medical Genetics, 2015, 16, 31.	2.1	26
38	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
39	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	1.3	24
40	Functional validation of novel compound heterozygous variants in B3GAT3 resulting in severe osteopenia and fractures: expanding the disease phenotype. BMC Medical Genetics, 2016, 17, 86.	2.1	22
41	Mutations in MECP2 exon 1 in classical rett patients disrupt MECP2_e1 transcription, but not transcription of MECP2_e2. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 210-216.	1.7	21
42	Pediatric Crohn Disease Presenting as Appendicitis: Differentiating Features from Typical Appendicitis. European Journal of Pediatric Surgery, 2012, 22, 274-278.	1.3	20
43	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
44	Variation among Consent Forms for Clinical Whole Exome Sequencing. Journal of Genetic Counseling, 2018, 27, 104-114.	1.6	18
45	Hypotonia and intellectual disability without dysmorphic features in a patient with PIGN-related disease. BMC Medical Genetics, 2017, 18, 124.	2.1	15
46	Challenges in genetic testing: clinician variant interpretation processes and the impact on clinical care. Genetics in Medicine, 2021, 23, 2289-2299.	2.4	15
47	Factors Affecting Migration to GRCh38 in Laboratories Performing Clinical Next-Generation Sequencing. Journal of Molecular Diagnostics, 2021, 23, 651-657.	2.8	13
48	Targeted next-generation sequencing revealed MYD88 deficiency in a child with chronic yersiniosis and granulomatous lymphadenitis. Journal of Allergy and Clinical Immunology, 2016, 137, 1591-1595.e4.	2.9	12
49	Arthrogryposis and pterygia as lethal end manifestations of genetically defined congenital myopathies. American Journal of Medical Genetics, Part A, 2018, 176, 359-367.	1.2	12
50	Allele Drop-Out in the MECP2 Gene Due to G-Quadruplex and i-Motif Sequences When Using Polymerase Chain Reaction-Based Diagnosis for Rett Syndrome. Genetic Testing and Molecular Biomarkers, 2010, 14, 241-247.	0.7	11
51	On the verge of diagnosis: Detection, reporting, and investigation of de novo variants in novel genes identified by clinical sequencing. Human Mutation, 2018, 39, 1505-1516.	2.5	9
52	Pathogenic variants in KPTN gene identified by clinical whole-genome sequencing. Journal of Physical Education and Sports Management, 2020, 6, a003970.	1.2	9
53	Two Brothers with Atypical UNC13D-Related Hemophagocytic Lymphohistiocytosis Characterized by Massive Lung and Brain Involvement. Frontiers in Immunology, 2017, 8, 1892.	4.8	8
54	De novo heterozygous missense and lossâ€ofâ€function variants in CDC42BPB are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973.	1.2	8

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55	High Molecular Diagnosis Rate in Undermasculinized Males with Differences in Sex Development Using a Stepwise Approach. Endocrinology, 2020, 161, .	2.8	7
56	The Future of Next-Generation Sequencing in Neurology. JAMA Neurology, 2015, 72, 971.	9.0	6
57	Novel heterozygous pathogenic variants in CHUK in a patient with AEC-like phenotype, immune deficiencies and 1q21.1 microdeletion syndrome: a case report. BMC Medical Genetics, 2018, 19, 41.	2.1	6
58	Nager syndrome in patient lacking acrofacial dysostosis: Expanding the phenotypic spectrum of <scp>SF3B4</scp>â€related disease. American Journal of Medical Genetics, Part A, 2021, 185, 1515-1518.	1.2	6
59	Clinical Stringency Greatly Improves Mutation Detection in Rett Syndrome. Canadian Journal of Neurological Sciences, 2005, 32, 321-326.	0.5	5
60	LZTR1 -Related Hypertrophic Cardiomyopathy Without Typical Noonan Syndrome Features. Circulation Genomic and Precision Medicine, 2020, 13, e002690.	3.6	5
61	Genotypeâ€“phenotype correlation in <scp>GNB1</scp>â€related neurodevelopmental disorder: Potential association of p.<scp>Leu95Pro</scp> with cleft palate. American Journal of Medical Genetics, Part A, 2021, 185, 1341-1343.	1.2	5
62	Clinical, Immunological, and Functional Characterization of Six Patients with Very High IgM Levels. Journal of Clinical Medicine, 2020, 9, 818.	2.4	4
63	Delayed diagnosis of holocarboxylase synthetase deficiency in three patients with prominent skin findings. Pediatric Dermatology, 2021, 38, 655-658.	0.9	4
64	Functionally impaired RPL8 variants associated with Diamondâ€“Blackfan anemia and a Diamondâ€“Blackfan anemiaâ€like phenotype. Human Mutation, 2022, 43, 389-402.	2.5	4
65	Big Data Strikes Again: Future Utilization of the UK Biobank as a Resource for Clinical Laboratories. Clinical Chemistry, 2021, 67, 932-934.	3.2	3
66	Phenotypic expansion and variable expressivity in individuals with JARID2â€related intellectual disability: A case series. Clinical Genetics, 2022, 102, 136-141.	2.0	3
67	Examination of rare genetic variants in dental enamel genes: The potential role of nextâ€generation sequencing in primary dental care. Orthodontics and Craniofacial Research, 2019, 22, 49-55.	2.8	1
68	An Unusual Presentation of Congenital Dyserythropoietic Anemia Type II (CDAII) Associated with Severe Anemia in a Patient with a Novel Mutation of the SEC23B Gene. Blood, 2012, 120, 990-990.	1.4	1
69	P-199â€ƒPathogenic CFTR Mutation in CrohnÊ¼s Disease in the Absence of Other CFTR-Related Manifestations. Inflammatory Bowel Diseases, 2016, 22, S69.	1.9	0
70	Classifying Germline Sequence Variants in the Era of Next-Generation Sequencing. Clinical Chemistry, 2016, 62, 799-806.	3.2	0
71	Coming up to Speed on Whole Genome Sequencing in Critically Ill Children. Advances in Molecular Pathology, 2018, 1, 1-8.	0.4	0
72	Second patient with GNB2-related neurodevelopmental disease: Further evidence for a gene-disease association. European Journal of Medical Genetics, 2021, 64, 104243.	1.3	0

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73	A novel likely-pathogenic variant in a patient with Hermansky-Pudlak Syndrome. Journal of Physical Education and Sports Management, 2021, 7, mcs.a006110.	1.2	0
74	Clinical Presentation of Polymerase E1 (POLE1) and Polymerase E2 (POLE2) Deficiencies. , 2019, , 1-6.		0
75	Clinical Presentation of Polymerase E1 (POLE1) and Polymerase E2 (POLE2) Deficiencies. , 2020, , 182-187.		0