John C Carey

List of Publications by Year
in descending order

135 papers	5,863 citations	81900 39 h-index	88630 70 g-index
149 all docs	149 docs citations	149 times ranked	6970 citing authors

#	Article	IF	Citations
1	Comprehensive variant calling from wholeâ€genome sequencing identifies a complex inversion that disrupts <scp>ZFPM2</scp> in familial congenital diaphragmatic hernia. Molecular Genetics & Genomic Medicine, 2022, 10, e1888.	1.2	6
2	Survival Outcomes of Infants with the Trisomy 13 or Trisomy 18 Syndromes. Journal of Pediatrics, 2022, 247, 11-13.	1.8	1
3	Maternal diabetesâ€related malformations in Utah : A population study of birth prevalence 2001â€“2016. Birth Defects Research, 2021, 113, 152-160.	1.5	1
4	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
5	Parentâ€authored memoirs: Lessons in the practice of narrative medicine. American Journal of Medical Genetics, Part A, 2021, 185, 2846-2848.	1.2	0
6	Natural history study of adults with <scp>Wolfâ€“Hirschhorn</scp> syndrome 1: Case series of personally observed 35 individuals. American Journal of Medical Genetics, Part A, 2021, 185, 1794-1802.	1.2	7
7	Parentâ€reported histories of adults with trisomy 13 syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1743-1756.	1.2	4
8	Thinking outside â€œThe Boxâ€• Caseâ€based didactics for medical education and the instructional legacy of Dr John M. Graham, Jr. American Journal of Medical Genetics, Part A, 2021, 185, 2636-2645.	1.2	3
9	Natural history study of adults with Wolfâ€“Hirschhorn syndrome 2: Patientâ€reported outcomes study. American Journal of Medical Genetics, Part A, 2021, 185, 2065-2069.	1.2	3
10	Reflections on observing faces in art. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 144-147.	1.6	1
11	The delineation of the <scp>Wolfâ€Hirschhorn</scp> syndrome over six decades: Illustration of the ongoing advances in phenotype analysis and cytogenomic technology. American Journal of Medical Genetics, Part A, 2021, 185, 2748-2755.	1.2	4
12	A celebration in honor of John M. Graham, Jr, <scp>MD</scp>, <scp>ScD</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 2617-2619.	1.2	0
13	Management of Children with the Trisomy 18 and Trisomy 13 Syndromes: Is there a Shift in the Paradigm of Care?. American Journal of Perinatology, 2021, 38, 1122-1125.	1.4	9
14	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
15	Emerging evidence that medical and surgical interventions improve the survival and outcome in the trisomy 13 and 18 syndromes. American Journal of Medical Genetics, Part A, 2020, 182, 13-14.	1.2	17
16	Exome Sequencing and Clinical Diagnosis. JAMA - Journal of the American Medical Association, 2020, 324, 627.	7.4	9
17	Experiences with offering pro bono medical genetics services in the West Indies: Benefits to patients, physicians, and the community. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1030-1041.	1.6	8
18	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25

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19	Application of exome sequencing to diagnose a novel presentation of the Cornelia de Lange syndrome in an Afroâ€Caribbean family. Molecular Genetics & Genomic Medicine, 2020, 8, e1318.	1.2	2
20	40th Annual David W Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2019 Annual Meeting. American Journal of Medical Genetics, Part A, 2020, 182, 877-942.	1.2	2
21	Exome Sequencing as Part of a Multidisciplinary Approach to Diagnosisâ€"Reply. JAMA - Journal of the American Medical Association, 2020, 324, 2445.	7.4	2
22	Novel de novo ARCN1 intronic variant causes rhizomelic short stature with microretrognathia and developmental delay. Journal of Physical Education and Sports Management, 2020, 6, a005728.	1.2	5
23	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
24	Solid tumor screening recommendations in trisomy 18. American Journal of Medical Genetics, Part A, 2019, 179, 455-466.	1.2	12
25	Mortality and Resource Use Following Cardiac Interventions in Children with Trisomy 13 and Trisomy 18 and Congenital Heart Disease. Pediatric Cardiology, 2019, 40, 349-356.	1.3	29
26	Osteoporosis and skeletal dysplasia caused by pathogenic variants in SGMS2. JCI Insight, 2019, 4, .	5.0	47
27	Three novel GJB2 (connexin 26) variants associated with autosomal dominant syndromic and nonsyndromic hearing loss. American Journal of Medical Genetics, Part A, 2018, 176, 945-950.	1.2	9
28	Deep phenotyping of patients with Tuberous Sclerosis Complex and no mutation identified in TSC1 and TSC2. European Journal of Medical Genetics, 2018, 61, 403-410.	1.3	25
29	A survey of antiepileptic drug responses identifies drugs with potential efficacy for seizure control in Wolfâ€"Hirschhorn syndrome. Epilepsy and Behavior, 2018, 81, 55-61.	1.7	14
30	Risk of hepatic neoplasms in Wolfâ€"Hirschhorn syndrome (4pâ€): Four new cases and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2389-2394.	1.2	11
31	Training Methods for Delivering Difficult News in Genetic Counseling and Genetics Residency Training Programs. Journal of Genetic Counseling, 2018, 27, 1497-1505.	1.6	4
32	M. Michael Cohen, Jr.: Author, diagnostician, geneticist, teacher, mentor, syndrome scholar extraordinaire (1937â€"2018). American Journal of Medical Genetics, Part A, 2018, 176, 1703-1705.	1.2	1
33	37th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2016 Annual Meeting. , 2017, 173, 2007-2073.		0
34	DDX3X mutations in two girls with a phenotype overlapping Toriolloâ€"Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373.	1.2	41
35	Introduction Special Series: Professor John M. Opitz, Founding Editor of AJMG, Awarded the Order of Merit from the Federal Republic of Germany. American Journal of Medical Genetics, Part A, 2017, 173, 1143-1144.	1.2	0
36	The spectrum of DNMT3A variants in Tattonâ€"Brownâ€"Rahman syndrome overlaps with that in hematologic malignancies. American Journal of Medical Genetics, Part A, 2017, 173, 3022-3028.	1.2	42

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37	Phenotype analysis of congenital and neurodevelopmental disorders in the next generation sequencing era. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 320-328.	1.6	5
38	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077.	12.8	72
39	Mycophenolate mofetil embryopathy: A newly recognized teratogenic syndrome. European Journal of Medical Genetics, 2017, 60, 16-21.	1.3	71
40	Etiology and clinical presentation of birth defects: population based study. BMJ: British Medical Journal, 2017, 357, j2249.	2.3	125
41	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. Molecular Autism, 2017, 8, 58.	4.9	37
42	Variable expressivity and incomplete penetrance in a large family with nonâ€classical Diamondâ€Blackfan anemia associated with ribosomal protein L11 splicing variant. American Journal of Medical Genetics, Part A, 2017, 173, 2622-2627.	1.2	14
43	Congenital Chylous Ascites and Ehlers-Danlos Syndrome Type VI. ACG Case Reports Journal, 2016, 3, e186.	0.4	1
44	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. PLoS Genetics, 2016, 12, e1006039.	3.5	18
45	Deletion 2q37 syndrome: Cognitiveâ€behavioral trajectories and autistic features related to breakpoint and deletion size. American Journal of Medical Genetics, Part A, 2016, 170, 2282-2291.	1.2	6
46	36th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2015 annual meeting. , 2016, 170, 1665-1726.		1
47	Clinical presentation and survival in a populationâ€based cohort of infants with gastroschisis in Utah, 1997â€“2011. American Journal of Medical Genetics, Part A, 2016, 170, 306-315.	1.2	19
48	Wilms tumor and trisomy 18: Is there an association?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 307-308.	1.6	11
49	Does medical intervention affect outcome in infants with trisomy 18 or trisomy 13?. American Journal of Medical Genetics, Part A, 2016, 170, 847-849.	1.2	16
50	Braddockâ€“Carey syndrome: A 21q22 contiguous gene syndrome encompassing RUNX1. American Journal of Medical Genetics, Part A, 2016, 170, 2580-2586.	1.2	12
51	Shared decision making and the pathways approach in the prenatal and postnatal management of the trisomy 13 and trisomy 18 syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 257-263.	1.6	27
52	Much ado about something 2: Reflections on the state of the American Journal of Medical Genetics 2016. American Journal of Medical Genetics, Part A, 2016, 170, 3067-3068.	1.2	0
53	Chromosomal microarray testing identifies a 4p terminal region associated with seizures in Wolfâ€“Hirschhorn syndrome. Journal of Medical Genetics, 2016, 53, 256-263.	3.2	40
54	Advances in the Understanding of the Genetic Causes of Hearing Loss in Children Inform a Rational Approach to Evaluation. Indian Journal of Pediatrics, 2016, 83, 1150-1156.	0.8	11

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55	Wolfâ€“Hirschhorn syndrome: A review and update. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 216-223.	1.6	119
56	Risk factors for Dandyâ€“Walker malformation: A populationâ€based assessment. American Journal of Medical Genetics, Part A, 2015, 167, 2009-2016.	1.2	14
57	Resistance strength training exercise in children with spinal muscular atrophy. Muscle and Nerve, 2015, 52, 559-567.	2.2	55
58	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. American Journal of Human Genetics, 2015, 96, 462-473.	6.2	124
59	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	6.2	55
60	Reflections on the etiology of structural birth defects: Established teratogens and risk factors. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 652-655.	1.6	13
61	Reported communication ability of persons with trisomy 18 and trisomy 13. Developmental Neurorehabilitation, 2015, 18, 322-329.	1.1	16
62	Assessment of Congenital Anomalies in Infants Born to Pregnant Women Enrolled in Clinical Trials. Clinical Infectious Diseases, 2014, 59, S428-S436.	5.8	12
63	Deletions involving genes WHSC1 and LETM1 may be necessary, but are not sufficient to cause Wolfâ€“Hirschhorn Syndrome. European Journal of Human Genetics, 2014, 22, 464-470.	2.8	43
64	Descriptive and risk factor analysis for choanal atresia: The National Birth Defects Prevention Study, 1997â€“2007. European Journal of Medical Genetics, 2014, 57, 220-229.	1.3	25
65	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	6.2	171
66	Confirmation of chromosomal microarray as a first-tier clinical diagnostic test for individuals with developmental delay, intellectual disability, autism spectrum disorders and dysmorphic features. European Journal of Paediatric Neurology, 2013, 17, 589-599.	1.6	170
67	Narrative medicine: A call to pens. American Journal of Medical Genetics, Part A, 2013, 161, 2117-2118.	1.2	3
68	Elements of morphology: General terms for congenital anomalies. American Journal of Medical Genetics, Part A, 2013, 161, 2726-2733.	1.2	101
69	Fine-grained facial phenotypeâ€“genotype analysis in Wolfâ€“Hirschhorn syndrome. European Journal of Human Genetics, 2012, 20, 33-40.	2.8	69
70	Perspectives on the care and management of infants with trisomy 18 and trisomy 13. Current Opinion in Pediatrics, 2012, 24, 672-678.	2.0	74
71	The trisomy 18 syndrome. Orphanet Journal of Rare Diseases, 2012, 7, 81.	2.7	269
72	Standard terminology for phenotypic variations: The Elements of Morphology project, its current progress, and future directions. Human Mutation, 2012, 33, 781-786.	2.5	24

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73	Analysis of skeletal dysplasias in the Utah population. American Journal of Medical Genetics, Part A, 2012, 158A, 1046-1054.	1.2	72
74	Response to Happle a novel X linked phenotype caused by hypomorphic EBP mutation. American Journal of Medical Genetics, Part A, 2011, 155, 1772-1772.	1.2	4
75	Abbreviations and terminology surrounding autism spectrum disorders and intellectual disability. American Journal of Medical Genetics, Part A, 2011, 155, 2905-2905.	1.2	2
76	Editorial comment: Foreword to very rare defects: What can we learn?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 251-251.	1.6	0
77	Reflections on mentoring. Developmental Dynamics, 2010, 239, 2521-2521.	1.8	0
78	The Importance of Case Reports in Advancing Scientific Knowledge of Rare Diseases. Advances in Experimental Medicine and Biology, 2010, 686, 77-86.	1.6	60
79	Elements of morphology: Introduction. American Journal of Medical Genetics, Part A, 2009, 149A, 2-5.	1.2	98
80	Preaxial hallucal polydactyly as a marker for diabetic embryopathy. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 13-19.	1.6	20
81	Spectrum of epilepsy and electroencephalogram patterns in Wolfâ€“Hirschhorn syndrome: experience with 87 patients. Developmental Medicine and Child Neurology, 2009, 51, 373-380.	2.1	68
82	Update on the clinical features and natural history of Wolfâ€“Hirschhorn (4pâ€) syndrome: Experience with 87 patients and recommendations for routine health supervision. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2008, 148C, 246-251.	1.6	150
83	Comprehensive analysis of Wolfâ€“Hirschhorn syndrome using array CGH indicates a high prevalence of translocations. European Journal of Human Genetics, 2008, 16, 45-52.	2.8	67
84	A multicenter study of the frequency and distribution of GJB2 and GJB6 mutations in a large North American cohort. Genetics in Medicine, 2007, 9, 413-426.	2.4	134
85	Introduction to hereditary deafness. American Journal of Medical Genetics, Part A, 2007, 143A, 1531-1532.	1.2	0
86	Two unique patients with novel microdeletions in 4p16.3 that exclude the WHS critical regions: Implications for critical region designation. American Journal of Medical Genetics, Part A, 2007, 143A, 2137-2142.	1.2	47
87	Introductory comments: M. Michael Cohen Jr. Festschrift. American Journal of Medical Genetics, Part A, 2007, 143A, 2851-2852.	1.2	2
88	A species not extinct: Publication of case reports and scientific knowledge. American Journal of Medical Genetics, Part A, 2006, 140A, 801-803.	1.2	14
89	Introductory comments special section: Trisomy 18. American Journal of Medical Genetics, Part A, 2006, 140A, 935-936.	1.2	8
90	Seizure and EEG patterns in Wolf-Hirschhorn (4p-) syndrome. Brain and Development, 2005, 27, 362-364.	1.1	30

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91	The methodology of the Utah Birth Defect Network: Congenital heart defects as an illustration. Birth Defects Research Part A: Clinical and Molecular Teratology, 2005, 73, 693-699.	1.6	26
92	Rubinstein-Taybi syndrome: New look at an â€œoldâ€•syndrome. American Journal of Medical Genetics Part A, 2005, 37, 1-2.	2.4	0
93	Three diagnostic signs in Williams syndrome. American Journal of Medical Genetics Part A, 2005, 37, 100-101.	2.4	11
94	Cornelia de Lange syndrome is caused by mutations in NIPBL, the human homolog of Drosophila melanogaster Nipped-B. Nature Genetics, 2004, 36, 631-635.	21.4	642
95	Status of the human malformation map: 2002. American Journal of Medical Genetics Part A, 2002, 115, 205-220.	2.4	4
96	Much ado about something: The place of theAmerican Journal of Medical Genetics in the field. American Journal of Medical Genetics Part A, 2001, 98, 1-2.	2.4	5
97	Exstrophy of the cloaca and the OEIS complex: One and the same. American Journal of Medical Genetics Part A, 2001, 99, 270-270.	2.4	46
98	Growth failure, intracranial calcifications, acquired pancytopenia, and unusual humoral immunodeficiency: A genetic syndrome?. American Journal of Medical Genetics Part A, 2000, 95, 17-20.	2.4	10
99	Diagnostic yield of the comprehensive assessment of developmental delay/mental retardation in an institute of child neuropsychiatry. , 1999, 82, 60-66.		102
100	Neurofibromatosis type 1: A model condition for the study of the molecular basis of variable expressivity in human disorders. American Journal of Medical Genetics Part A, 1999, 89, 7-13.	2.4	66
101	Microcephaly with simplified gyral pattern in six related children. , 1999, 84, 137-144.		26
102	Descriptive analysis of tibial pseudarthrosis in patients with neurofibromatosis 1. , 1999, 84, 413-419.		114
103	Polytopic anomalies with agenesis of the lower vertebral column. American Journal of Medical Genetics Part A, 1999, 87, 99-114.	2.4	61
104	Natural History of Wolf-Hirschhorn Syndrome: Experience With 15 Cases. Pediatrics, 1999, 103, 830-836.	2.1	124
105	Variegated aneuploidy in two siblings: Phenotype, genotype, CENP-E analysis, and literature review. , 1998, 75, 45-51.		13
106	Neurofibromatosis-Noonan syndrome. American Journal of Medical Genetics Part A, 1998, 75, 263-264.	2.4	39
107	Wolf-Hirschhorn syndrome and Pitt-Rogers-Danks syndrome. , 1998, 75, 541-541.		22
108	Wolfâ€Hirschhorn syndrome and Pittâ€Rogersâ€Danks syndrome. American Journal of Medical Genetics Part A, 1998, 75, 541-541.	2.4	1

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109	Six patients with oral-facial-digital syndrome IV: The case for heterogeneity. American Journal of Medical Genetics Part A, 1997, 69, 250-260.	2.4	23
110	Umbilical cord agenesis in limb body wall defect. , 1997, 71, 97-105.		27
111	Xq28-linked noncompaction of the left ventricular myocardium: Prenatal diagnosis and pathologic analysis of affected individuals. American Journal of Medical Genetics Part A, 1997, 72, 257-265.	2.4	254
112	Six patients with oralâ€facialâ€digital syndrome IV: The case for heterogeneity. American Journal of Medical Genetics Part A, 1997, 69, 250-260.	2.4	1
113	Xq28â€linked noncompaction of the left ventricular myocardium: Prenatal diagnosis and pathologic analysis of affected individuals. American Journal of Medical Genetics Part A, 1997, 72, 257-265.	2.4	15
114	Fibroblast growth factor receptor 2 mutations in Beareâ€“Stevenson cutis gyrata syndrome. Nature Genetics, 1996, 13, 492-494.	21.4	181
115	Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival, and recurrence risk. American Journal of Medical Genetics Part A, 1994, 49, 175-188.	2.4	252
116	Natural history of trisomy 18 and trisomy 13: II. Psychomotor development. American Journal of Medical Genetics Part A, 1994, 49, 189-194.	2.4	111
117	Pulmonary hyperplasia in the Fraser cryptophthalmos syndrome. American Journal of Medical Genetics Part A, 1994, 52, 427-431.	2.4	20
118	Brachymesomelia and Peters anomaly: A new syndrome. American Journal of Medical Genetics Part A, 1993, 45, 416-419.	2.4	11
119	Radiological features in Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1006-1013.	2.4	44
120	Nosological Considerations of the Neurofibromatoses. Journal of Dermatology, 1992, 19, 873-880.	1.2	18
121	New syndrome involving the visual, auditory, respiratory, gastrointestinal, and renal systems. American Journal of Medical Genetics Part A, 1992, 44, 461-464.	2.4	1
122	New syndrome? Osteochondrodysplasia with rhizomelia, platyspondyly, callosal agenesis, thrombocytopenia, hydrocephalus, and hypertension. American Journal of Medical Genetics Part A, 1991, 40, 183-187.	2.4	7
123	The Otto Ullrich award for excellence in clinical genetics. American Journal of Medical Genetics Part A, 1991, 41, 125-125.	2.4	1
124	Prader Willi scientific symposium Salt Lake City, Utah July 18, 1990. American Journal of Medical Genetics Part A, 1991, 41, 524-530.	2.4	2
125	Critical review of articles regarding pregnancy exposures in popular magazines. Teratology, 1990, 42, 469-472.	1.6	14
126	Congenital hypoplastic (Diamond-Blackfan) anemia in seven members of one kindred. American Journal of Medical Genetics Part A, 1990, 35, 251-256.	2.4	30

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127	Thoracic Volume Reduction as a Mechanism for Pulmonary Hypoplasia in Chondrodystrophic Mice. Pediatric Pathology, 1990, 10, 919-929.	0.5	10
128	Rubinstein-Taybi syndrome: A natural history study. American Journal of Medical Genetics Part A, 1990, 37, 30-37.	2.4	52
129	Di George Anomaly and Velocardiofacial Syndrome. Pediatrics, 1990, 85, 526-530.	2.1	48
130	Pulmonary Hypoplasia in Mice Homozygous for the Cartilage Matrix Deficiency (Cmd) Gene: A Model for a Human Congenital Disorder. Pediatric Pathology, 1989, 9, 501-512.	0.5	10
131	Corpus callosum agenesis, facial anomalies, Robin sequence, and other anomalies: A new autosomal recessive syndrome?. American Journal of Medical Genetics Part A, 1988, 31, 17-23.	2.4	69
132	Chondrodystrophic mice with coincidental agnathia: Evidence for the tongue obstruction hypothesis in cleft palate. Teratology, 1988, 38, 565-570.	1.6	22
133	Delineation of the male phenotype in craniofrontonasal syndrome. American Journal of Medical Genetics Part A, 1987, 27, 623-631.	2.4	43
134	Linkage analysis of Von Recklinghausen neurofibromatosis: Chromosomes 4 and 19. Genetic Epidemiology, 1986, 3, 313-321.	1.3	7
135	Confirmation of the Cohen syndrome. Journal of Pediatrics, 1978, 93, 239-244.	1.8	72