John C Carey

List of Publications by Year
in descending order

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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	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
2	The trisomy 18 syndrome. Orphanet Journal of Rare Diseases, 2012, 7, 81.	2.7	269
3	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
4	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
5	Fibroblast growth factor receptor 2 mutations in Beareâ€“Stevenson cutis gyrata syndrome. Nature Genetics, 1996, 13, 492-494.	21.4	181
6	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
7	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
8	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
9	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
10	Etiology and clinical presentation of birth defects: population based study. BMJ: British Medical Journal, 2017, 357, j2249.	2.3	125
11	Natural History of Wolf-Hirschhorn Syndrome: Experience With 15 Cases. Pediatrics, 1999, 103, 830-836.	2.1	124
12	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
13	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
14	Descriptive analysis of tibial pseudarthrosis in patients with neurofibromatosis 1. , 1999, 84, 413-419.		114
15	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
16	Diagnostic yield of the comprehensive assessment of developmental delay/mental retardation in an institute of child neuropsychiatry. , 1999, 82, 60-66.		102
17	Elements of morphology: General terms for congenital anomalies. American Journal of Medical Genetics, Part A, 2013, 161, 2726-2733.	1.2	101
18	Elements of morphology: Introduction. American Journal of Medical Genetics, Part A, 2009, 149A, 2-5.	1.2	98

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19	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
20	Confirmation of the Cohen syndrome. Journal of Pediatrics, 1978, 93, 239-244.	1.8	72
21	Analysis of skeletal dysplasias in the Utah population. American Journal of Medical Genetics, Part A, 2012, 158A, 1046-1054.	1.2	72
22	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077.	12.8	72
23	Mycophenolate mofetil embryopathy: A newly recognized teratogenic syndrome. European Journal of Medical Genetics, 2017, 60, 16-21.	1.3	71
24	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
25	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
26	Fine-grained facial phenotypeâ€“genotype analysis in Wolfâ€“Hirschhorn syndrome. European Journal of Human Genetics, 2012, 20, 33-40.	2.8	69
27	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
28	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
29	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
30	Polytopic anomalies with agenesis of the lower vertebral column. American Journal of Medical Genetics Part A, 1999, 87, 99-114.	2.4	61
31	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
32	Resistance strength training exercise in children with spinal muscular atrophy. Muscle and Nerve, 2015, 52, 559-567.	2.2	55
33	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	6.2	55
34	Rubinstein-Taybi syndrome: A natural history study. American Journal of Medical Genetics Part A, 1990, 37, 30-37.	2.4	52
35	Di George Anomaly and Velocardiofacial Syndrome. Pediatrics, 1990, 85, 526-530.	2.1	48
36	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

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37	Osteoporosis and skeletal dysplasia caused by pathogenic variants in SGMS2. JCI Insight, 2019, 4, .	5.0	47
38	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
39	Radiological features in Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1993, 47, 1006-1013.	2.4	44
40	Delineation of the male phenotype in craniofrontonasal syndrome. American Journal of Medical Genetics Part A, 1987, 27, 623-631.	2.4	43
41	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
42	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
43	DDX3X mutations in two girls with a phenotype overlapping Torielloâ€“Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373.	1.2	41
44	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
45	Neurofibromatosis-Noonan syndrome. American Journal of Medical Genetics Part A, 1998, 75, 263-264.	2.4	39
46	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. Molecular Autism, 2017, 8, 58.	4.9	37
47	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
48	Seizure and EEG patterns in Wolf-Hirschhorn (4p-) syndrome. Brain and Development, 2005, 27, 362-364.	1.1	30
49	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
50	Umbilical cord agenesis in limb body wall defect. , 1997, 71, 97-105.		27
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	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
53	Microcephaly with simplified gyral pattern in six related children. , 1999, 84, 137-144.		26
54	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

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55	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
56	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
57	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
58	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
59	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
60	Chondrodystrophic mice with coincidental agnathia: Evidence for the tongue obstruction hypothesis in cleft palate. Teratology, 1988, 38, 565-570.	1.6	22
61	Wolf-Hirschhorn syndrome and Pitt-Rogers-Danks syndrome. , 1998, 75, 541-541.		22
62	Pulmonary hyperplasia in the Fraser cryptophthalmos syndrome. American Journal of Medical Genetics Part A, 1994, 52, 427-431.	2.4	20
63	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
64	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
65	Nosological Considerations of the Neurofibromatoses. Journal of Dermatology, 1992, 19, 873-880.	1.2	18
66	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. PLoS Genetics, 2016, 12, e1006039.	3.5	18
67	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
68	Reported communication ability of persons with trisomy 18 and trisomy 13. Developmental Neurorehabilitation, 2015, 18, 322-329.	1.1	16
69	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
70	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
71	Critical review of articles regarding pregnancy exposures in popular magazines. Teratology, 1990, 42, 469-472.	1.6	14
72	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

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73	Risk factors for Dandyâ€“Walker malformation: A populationâ€based assessment. American Journal of Medical Genetics, Part A, 2015, 167, 2009-2016.	1.2	14
74	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
75	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
76	Variegated aneuploidy in two siblings: Phenotype, genotype, CENP-E analysis, and literature review. , 1998, 75, 45-51.		13
77	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
78	Assessment of Congenital Anomalies in Infants Born to Pregnant Women Enrolled in Clinical Trials. Clinical Infectious Diseases, 2014, 59, S428-S436.	5.8	12
79	Braddockâ€“Carey syndrome: A 21q22 contiguous gene syndrome encompassing RUNX1. American Journal of Medical Genetics, Part A, 2016, 170, 2580-2586.	1.2	12
80	Solid tumor screening recommendations in trisomy 18. American Journal of Medical Genetics, Part A, 2019, 179, 455-466.	1.2	12
81	Brachymesomelia and Peters anomaly: A new syndrome. American Journal of Medical Genetics Part A, 1993, 45, 416-419.	2.4	11
82	Three diagnostic signs in Williams syndrome. American Journal of Medical Genetics Part A, 2005, 37, 100-101.	2.4	11
83	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
84	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
85	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
86	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
87	Thoracic Volume Reduction as a Mechanism for Pulmonary Hypoplasia in Chondrodystrophic Mice. Pediatric Pathology, 1990, 10, 919-929.	0.5	10
88	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
89	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
90	Exome Sequencing and Clinical Diagnosis. JAMA - Journal of the American Medical Association, 2020, 324, 627.	7.4	9

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91	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
92	Introductory comments special section: Trisomy 18. American Journal of Medical Genetics, Part A, 2006, 140A, 935-936.	1.2	8
93	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
94	Linkage analysis of Von Recklinghausen neurofibromatosis: Chromosomes 4 and 19. Genetic Epidemiology, 1986, 3, 313-321.	1.3	7
95	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
96	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
97	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
98	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
99	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
100	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
101	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
102	Status of the human malformation map: 2002. American Journal of Medical Genetics Part A, 2002, 115, 205-220.	2.4	4
103	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
104	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
105	Parentâ€reported histories of adults with trisomy 13 syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1743-1756.	1.2	4
106	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
107	Narrative medicine: A call to pens. American Journal of Medical Genetics, Part A, 2013, 161, 2117-2118.	1.2	3
108	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

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109	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
110	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
111	Introductory comments: M. Michael Cohen Jr. Festschrift. American Journal of Medical Genetics, Part A, 2007, 143A, 2851-2852.	1.2	2
112	Abbreviations and terminology surrounding autism spectrum disorders and intellectual disability. American Journal of Medical Genetics, Part A, 2011, 155, 2905-2905.	1.2	2
113	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
114	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
115	Exome Sequencing as Part of a Multidisciplinary Approach to Diagnosisâ€”Reply. JAMA - Journal of the American Medical Association, 2020, 324, 2445.	7.4	2
116	The Otto Ullrich award for excellence in clinical genetics. American Journal of Medical Genetics Part A, 1991, 41, 125-125.	2.4	1
117	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
118	Congenital Chylous Ascites and Ehlers-Danlos Syndrome Type VI. ACG Case Reports Journal, 2016, 3, e186.	0.4	1
119	36th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2015 annual meeting. , 2016, 170, 1665-1726.		1
120	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
121	Maternal diabetesâ€related malformations in Utah : A population study of birth prevalence 2001â€“2016. Birth Defects Research, 2021, 113, 152-160.	1.5	1
122	Reflections on observing faces in art. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 144-147.	1.6	1
123	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
124	Wolfâ€Hirschhorn syndrome and Pittâ€Rogersâ€Danks syndrome. American Journal of Medical Genetics Part A, 1998, 75, 541-541.	2.4	1
125	Survival Outcomes of Infants with the Trisomy 13 or Trisomy 18 Syndromes. Journal of Pediatrics, 2022, 247, 11-13.	1.8	1
126	Rubinstein-Taybi syndrome: New look at an â€œoldâ€•syndrome. American Journal of Medical Genetics Part A, 2005, 37, 1-2.	2.4	0

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127	Introduction to hereditary deafness. American Journal of Medical Genetics, Part A, 2007, 143A, 1531-1532.	1.2	0
128	Reflections on mentoring. Developmental Dynamics, 2010, 239, 2521-2521.	1.8	0
129	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
130	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
131	37th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2016 Annual Meeting. , 2017, 173, 2007-2073.		0
132	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
133	Parentâ€authored memoirs: Lessons in the practice of narrative medicine. American Journal of Medical Genetics, Part A, 2021, 185, 2846-2848.	1.2	0
134	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
135	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0