Albert E Chudley

List of Publications by Year
in descending order

103 papers	6,316 citations	94433 37 h-index	71685 76 g-index
106 all docs	106 docs citations	106 times ranked	6965 citing authors

#	Article	IF	Citations
1	Mutations in the Gene for Cardiac Myosin-Binding Protein C and Late-Onset Familial Hypertrophic Cardiomyopathy. New England Journal of Medicine, 1998, 338, 1248-1257.	27.0	701
2	Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis. Cmaj, 2005, 172, S1-S21.	2.0	701
3	Fetal alcohol spectrum disorder: a guideline for diagnosis across the lifespan. Cmaj, 2016, 188, 191-197.	2.0	379
4	Comorbidity of fetal alcohol spectrum disorder: a systematic review and meta-analysis. Lancet, The, 2016, 387, 978-987.	13.7	368
5	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215
6	Homozygous Deletion of the Very Low Density Lipoprotein Receptor Gene Causes Autosomal Recessive Cerebellar Hypoplasia with Cerebral Gyral Simplification. American Journal of Human Genetics, 2005, 77, 477-483.	6.2	192
7	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 2011, 89, 713-730.	6.2	178
8	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	6.2	171
9	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	6.2	157
10	Intragenic loss of function mutations demonstrate the primary role of FMR1 in fragile X syndrome. Nature Genetics, 1995, 10, 483-485.	21.4	152
11	Kabuki syndrome: international consensus diagnostic criteria. Journal of Medical Genetics, 2019, 56, 89-95.	3.2	146
12	DNA methylation signature of human fetal alcohol spectrum disorder. Epigenetics and Chromatin, 2016, 9, 25.	3.9	129
13	CODAS Syndrome Is Associated with Mutations of LONP1, Encoding Mitochondrial AAA+ Lon Protease. American Journal of Human Genetics, 2015, 96, 121-135.	6.2	127
14	Fragile X syndrome. Journal of Pediatrics, 1987, 110, 821-831.	1.8	116
15	GPSM2 Mutations Cause the Brain Malformations and Hearing Loss in Chudley-McCullough Syndrome. American Journal of Human Genetics, 2012, 90, 1088-1093.	6.2	103
16	Evaluation of Spatial Working Memory Function in Children and Adults with Fetal Alcohol Spectrum Disorders: A Functional Magnetic Resonance Imaging Study. Pediatric Research, 2005, 58, 1150-1157.	2.3	100
17	DNA methylation as a predictor of fetal alcohol spectrum disorder. Clinical Epigenetics, 2018, 10, 5.	4.1	89
18	Relationships between Head Circumference, Brain Volume and Cognition in Children with Prenatal Alcohol Exposure. PLoS ONE, 2016, 11, e0150370.	2.5	87

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19	Genetic evaluation of pervasive developmental disorders: the terminal 22q13 deletion syndrome may represent a recognizable phenotype. Clinical Genetics, 2000, 57, 103-109.	2.0	82
20	Population-based prevalence of fetal alcohol spectrum disorder in Canada. BMC Public Health, 2019, 19, 845.	2.9	79
21	Fragile (X) X-linked mental retardation I: Relationship between age and intelligence and the frequency of expression of fragil (X)(q28). American Journal of Medical Genetics Part A, 1983, 14, 699-712.	2.4	77
22	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	6.2	77
23	The DLX1and DLX2 genes and susceptibility to autism spectrum disorders. European Journal of Human Genetics, 2009, 17, 228-235.	2.8	75
24	Subtelomeric deletions of chromosome 6p: Molecular and cytogenetic characterization of three new cases with phenotypic overlap with Ritscher?Schinzel (3C) syndrome. American Journal of Medical Genetics, Part A, 2005, 134A, 3-11.	1.2	70
25	Successful pregnancy following continuous treatment with combination chemotherapy before conception and throughout pregnancy. Cancer, 1984, 54, 800-803.	4.1	69
26	Association of GTF2i in the Williams-Beuren Syndrome Critical Region with Autism Spectrum Disorders. Journal of Autism and Developmental Disorders, 2012, 42, 1459-1469.	2.7	61
27	Manitoba-oculo-tricho-anal (MOTA) syndrome is caused by mutations in FREM1. Journal of Medical Genetics, 2011, 48, 375-382.	3.2	60
28	A novel mutation inKIAA0196: identification of a gene involved in Ritscherâ€“Schinzel/3C syndrome in a First Nations cohort. Journal of Medical Genetics, 2013, 50, 819-822.	3.2	60
29	Outcomes of Genetic Evaluation in Children with Pervasive Developmental Disorder. Journal of Developmental and Behavioral Pediatrics, 1998, 19, 321-325.	1.1	56
30	Autism in fragile X females. American Journal of Medical Genetics Part A, 1986, 23, 375-380.	2.4	55
31	Human Brain Abnormalities Associated With Prenatal Alcohol Exposure and Fetal Alcohol Spectrum Disorder. Journal of Neuropathology and Experimental Neurology, 2017, 76, 813-833.	1.7	55
32	Mutation in the 5â€² alternatively spliced region of the XNP/ATR-X gene causes Chudleyâ€“Lowry syndrome. European Journal of Human Genetics, 2005, 13, 176-183.	2.8	53
33	Itâ€™s a Shame! Stigma Against Fetal Alcohol Spectrum Disorder: Examining the Ethical Implications for Public Health Practices and Policies. Public Health Ethics, 2016, 9, 65-77.	1.0	50
34	Agenesis of the penis: Patterns of associated malformations. , 1999, 84, 47-55.		49
35	Cost of Fetal Alcohol Spectrum Disorder Diagnosis in Canada. PLoS ONE, 2013, 8, e60434.	2.5	49
36	Identifying fetal alcohol spectrum disorder in primary care. Cmaj, 2005, 172, 628-630.	2.0	44

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37	Challenges of diagnosis in fetal alcohol syndrome and fetal alcohol spectrum disorder in the adult. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 261-272.	1.6	44
38	Clinical genetics and the Hutterite population: A review of Mendelian disorders. American Journal of Medical Genetics, Part A, 2008, 146A, 1088-1098.	1.2	44
39	Bilateral sensorineural deafness and hydrocephalus due to foramen of Monro obstruction in sibs: A newly described autosomal recessive disorder. , 1997, 68, 350-356.		38
40	Unique disease heritage of the Dutchâ€German Mennonite population. American Journal of Medical Genetics, Part A, 2008, 146A, 1072-1087.	1.2	38
41	Overview of the Genetic Basis and Epigenetic Mechanisms that Contribute to FASD Pathobiology. Current Topics in Medicinal Chemistry, 2017, 17, 808-828.	2.1	38
42	Mental retardation, distinct facial changes, short stature, obesity, and hypogonadism: A new X-linked mental retardation syndrome. American Journal of Medical Genetics Part A, 1988, 31, 741-751.	2.4	36
43	Comparison of spatial working memory in children with prenatal alcohol exposure and those diagnosed with ADHD; A functional magnetic resonance imaging study. Journal of Neurodevelopmental Disorders, 2012, 4, 12.	3.1	36
44	Zimmerman-Laband syndrome and profound mental retardation. American Journal of Medical Genetics Part A, 1986, 25, 543-547.	2.4	34
45	Autosomal recessive cerebellar hypoplasia in the Hutterite population. Developmental Medicine and Child Neurology, 2005, 47, 691.	2.1	34
46	Evidence for Ritscherâ€Schinzel syndrome in Canadian native Indians. American Journal of Medical Genetics Part A, 1995, 56, 343-350.	2.4	33
47	Fetal alcohol spectrum disorder: counting the invisible - mission impossible?. Archives of Disease in Childhood, 2008, 93, 721-722.	1.9	31
48	Prevalence of Pervasive Developmental Disorders in Two Canadian Provinces. Journal of Policy and Practice in Intellectual Disabilities, 2006, 3, 164-172.	2.7	30
49	2p15â€“p16.1 microdeletion syndrome: molecular characterization and association of the OTX1 and XPO1 genes with autism spectrum disorders. European Journal of Human Genetics, 2011, 19, 1264-1270.	2.8	30
50	SAMS, a Syndrome of Short Stature, Auditory-Canal Atresia, Mandibular Hypoplasia, and Skeletal Abnormalities Is a Unique Neurocristopathy Caused by Mutations in Goosecoid. American Journal of Human Genetics, 2013, 93, 1135-1142.	6.2	30
51	New familial syndrome of unilateral upper eyelid coloboma, aberrant anterior hairline pattern, and anal anomalies in Manitoba Indians. American Journal of Medical Genetics Part A, 1992, 42, 793-799.	2.4	29
52	Newly recognized syndrome of cerebral, ocular, dental, auricular, skeletal anomalies: CODAS syndromeâ€"a case report. American Journal of Medical Genetics Part A, 1991, 40, 88-93.	2.4	27
53	Tibial agenesis, femoral duplication, and caudal midline anomalies. American Journal of Medical Genetics Part A, 1999, 85, 13-19.	2.4	25
54	Familial duodenal atresia: A report of two families and review. American Journal of Medical Genetics Part A, 1989, 34, 442-444.	2.4	23

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55	Visual search for feature conjunctions: an fMRI study comparing alcohol-related neurodevelopmental disorder (ARND) to ADHD. Journal of Neurodevelopmental Disorders, 2015, 7, 10.	3.1	23
56	Floating-Harbor syndrome and celiac disease. American Journal of Medical Genetics Part A, 1991, 38, 562-564.	2.4	22
57	True precocious puberty in a girl with the fragile X syndrome. American Journal of Medical Genetics Part A, 1990, 37, 265-267.	2.4	20
58	Manitoba oculotrichoanal (MOTA) syndrome: Report of eight new cases. American Journal of Medical Genetics, Part A, 2007, 143A, 853-857.	1.2	19
59	Functional Evaluation of Hidden Figures Object Analysis in Children with Autistic Disorder. Journal of Autism and Developmental Disorders, 2011, 41, 13-22.	2.7	19
60	The greig cephalopolysyndactyly syndrome in a canadian family. American Journal of Medical Genetics Part A, 1982, 13, 269-276.	2.4	18
61	Facial weakness and oligosyndactyly: ? independent variable features of familial type of the MÃ¶bius syndrome. Clinical Genetics, 1983, 24, 350-354.	2.0	18
62	Fra(2) (q13) and inv(9) (pllq12) in autism: Causal relationship?. American Journal of Medical Genetics Part A, 1986, 23, 381-392.	2.4	18
63	Proximal interstitial 6q deletion: A recognizable syndrome. American Journal of Medical Genetics Part A, 1997, 71, 353-356.	2.4	18
64	Fetal Alcohol Spectrum Disorder. , 2012, , 443-452.		18
65	Ring chromosome 17 in a mentally retarded young manâ€"clinical, cytogenetic, and biochemical investigations. American Journal of Medical Genetics Part A, 1982, 12, 219-225.	2.4	17
66	Third case of cerebral, ocular, dental, auricular, skeletal anomalies (CODAS) syndrome, further delineating a new malformation syndrome: First report of an affected male and review of literature. American Journal of Medical Genetics Part A, 2001, 102, 44-47.	2.4	17
67	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	1.2	17
68	Meeting the needs of future physicians: a core curriculum initiative for postgraduate medical education at a Canadian university. Medical Education, 2001, 35, 973-982.	2.1	16
69	Development of Canadian screening tools for fetal alcohol spectrum disorder. Journal of Population Therapeutics and Clinical Pharmacology, 2008, 15, e344-66.	1.9	16
70	Copy number variation in fetal alcohol spectrum disorder. Biochemistry and Cell Biology, 2018, 96, 161-166.	2.0	15
71	Short-term memory and cognitive variability in adult fragile X females. American Journal of Medical Genetics Part A, 1991, 38, 488-492.	2.4	14
72	Multicore disease in sibs with severe mental retardation, short stature, facial anomalies, hypoplasia of the pituitary fossa, and hypogonadotrophic hypogonadism. American Journal of Medical Genetics Part A, 1985, 20, 145-158.	2.4	13

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73	Recognizable behavioral and somatic phenotype in patients with proximal interstitial 18q deletion: Report on a new affected child and follow-up on the original reported familial cases. American Journal of Medical Genetics Part A, 1992, 43, 535-538.	2.4	13
74	Diagnosis of fetal alcohol spectrum disorder: current practices and future considerations. Biochemistry and Cell Biology, 2018, 96, 231-236.	2.0	13
75	Familial supernumerary microchromosome mosaicism: Phenotypic effects and an attempt at characterization. American Journal of Medical Genetics Part A, 1983, 16, 89-97.	2.4	12
76	Mucinous cystadenoma of ovary in a patient with Williams syndrome. American Journal of Medical Genetics Part A, 1993, 46, 349-349.	2.4	12
77	X-linked mental retardation syndrome with seizures, hypogammaglobulinemia, and progressive gait disturbance is regionally mapped between Xq21.33 and Xq23. , 1999, 85, 255-262.		12
78	Vertical transmission of the Ohdo blepharophimosis syndrome. American Journal of Medical Genetics Part A, 1998, 77, 144-148.	2.4	10
79	Fetal hepatic haemangioendothelioma: a new association with elevated maternal serum alpha-fetoprotein. , 2000, 20, 432-434.		8
80	Ulnar agenesis and endocardial fibroelastosis. American Journal of Medical Genetics Part A, 1990, 37, 258-260.	2.4	7
81	Segregation analysis of rare autosomal folate sensitive fragile sites. American Journal of Medical Genetics Part A, 1993, 46, 165-171.	2.4	7
82	SAMS: Provisionally unique multiple congenital anomalies syndrome consisting of short stature, auditory canal atresia, mandibular hypoplasia, and skeletal abnormalities. , 1998, 75, 256-260.		7
83	Possible association of rare autosomal folate sensitive fragile sites and idiopathic mental retardation: a blind controlled population study. Clinical Genetics, 1990, 38, 241-256.	2.0	7
84	A Case of an Infant With Compound Heterozygous Mutations for Hypertrophic Cardiomyopathy Producing a Phenotype of Left Ventricular Noncompaction. Canadian Journal of Cardiology, 2014, 30, 1249.e1-1249.e3.	1.7	7
85	Developmental delay, short stature, and minor facial anomalies in a child with ring chromosome 16. American Journal of Medical Genetics Part A, 1988, 31, 145-151.	2.4	6
86	A notâ€soâ€â€œnewâ€•mental retardation syndrome. American Journal of Medical Genetics Part A, 2002, 111, 106-106.	2.4	6
87	Possible prenatal hydantoin effect in a child born to a nonepileptic mother. American Journal of Medical Genetics Part A, 1987, 27, 373-378.	2.4	5
88	Radiographic characterization of the hands in Ritscher-Schinzel/3-C syndrome. SpringerPlus, 2013, 2, 594.	1.2	5
89	Meeting the needs of future physicians: a core curriculum initiative for postgraduate medical education at a Canadian university. Medical Education, 2001, 35, 973-982.	2.1	4
90	Lissencephaly With Brainstem and Cerebellar Hypoplasia and Congenital Cataracts. Journal of Child Neurology, 2014, 29, 860-864.	1.4	3

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91	Fetal Alcohol Spectrum Disorderâ€"High Rates, High Needs, High Time for Action. JAMA Pediatrics, 2017, 171, 940.	6.2	3
92	Classifications of split hand foot malformation (SHFM) should include transverse deficiencies: Why Maisels was correct. American Journal of Medical Genetics, Part A, 2021, 185, 2809-2814.	1.2	3
93	Genetic landmarks through philately - Henry Louis â€˜Louâ€™ Gehrig and amyotrophic lateral sclerosis. Clinical Genetics, 1999, 56, 425-427.	2.0	2
94	Letters to the Editor. Journal of Developmental and Behavioral Pediatrics, 1999, 20, 72-73.	1.1	2
95	History of genetics through philately - Carl Linnaeus (Carl von LinnÃ©). Clinical Genetics, 2008, 60, 104-106.	2.0	2
96	Effects of in vivo diagnostic ultrasound on SCE frequency in cultured amniocytes. American Journal of Medical Genetics Part A, 1982, 13, 349-350.	2.4	1
97	Autosomal recessive cerebellar hypoplasia in the Hutterite population. Developmental Medicine and Child Neurology, 2007, 47, 691-695.	2.1	1
98	Orthopaedic Aspects of SAMS Syndrome. Journal of Pediatric Genetics, 2022, 11, 051-058.	0.7	1
99	Response to "A critique for the new Canadian FASD diagnostic Guidelines". Journal of the Canadian Academy of Child and Adolescent Psychiatry, 2018, 27, 83-87.	0.6	1
100	Genetic landmarks through philately - The Habsburg jaw. Clinical Genetics, 2008, 54, 283-284.	2.0	0
101	FETAL ALCOHOL SPECTRUM DISORDER: COST OF SCREENING AND DIAGNOSIS IN CANADA. Journal of Epidemiology and Community Health, 2013, 67, e2.20-e2.	3.7	0
102	Response to Correspondence on â€œLissencephaly With Brainstem and Cerebellar Hypoplasia and Congenital Cataractsâ€. Journal of Child Neurology, 2015, 30, 666-666.	1.4	0
103	Teratogenic Influences on Cerebellar Development. , 2017, , 275-300.		0