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	Orthopaedic Aspects of SAMS Syndrome. Journal of Pediatric Genetics, 2022, 11, 051-058.	0.7	1
2	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
3	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
4	Population-based prevalence of fetal alcohol spectrum disorder in Canada. BMC Public Health, 2019, 19, 845.	2.9	79
5	Kabuki syndrome: international consensus diagnostic criteria. Journal of Medical Genetics, 2019, 56, 89-95.	3.2	146
6	Copy number variation in fetal alcohol spectrum disorder. Biochemistry and Cell Biology, 2018, 96, 161-166.	2.0	15
7	Diagnosis of fetal alcohol spectrum disorder: current practices and future considerations. Biochemistry and Cell Biology, 2018, 96, 231-236.	2.0	13
8	DNA methylation as a predictor of fetal alcohol spectrum disorder. Clinical Epigenetics, 2018, 10, 5.	4.1	89
9	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
10	Fetal Alcohol Spectrum Disorderâ€"High Rates, High Needs, High Time for Action. JAMA Pediatrics, 2017, 171, 940.	6.2	3
11	Teratogenic Influences on Cerebellar Development. , 2017, , 275-300.		0
12	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
13	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
14	DNA methylation signature of human fetal alcohol spectrum disorder. Epigenetics and Chromatin, 2016, 9, 25.	3.9	129
15	Fetal alcohol spectrum disorder: a guideline for diagnosis across the lifespan. Cmaj, 2016, 188, 191-197.	2.0	379
16	Comorbidity of fetal alcohol spectrum disorder: a systematic review and meta-analysis. Lancet, The, 2016, 387, 978-987.	13.7	368
17	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
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	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
20	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
21	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215
22	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
23	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
24	Lissencephaly With Brainstem and Cerebellar Hypoplasia and Congenital Cataracts. Journal of Child Neurology, 2014, 29, 860-864.	1.4	3
25	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
26	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
27	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
28	Radiographic characterization of the hands in Ritscher-Schinzel/3-C syndrome. SpringerPlus, 2013, 2, 594.	1.2	5
29	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
30	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
31	Cost of Fetal Alcohol Spectrum Disorder Diagnosis in Canada. PLoS ONE, 2013, 8, e60434.	2.5	49
32	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
33	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
34	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
35	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
36	Fetal Alcohol Spectrum Disorder. , 2012, , 443-452.		18

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37	Manitoba-oculo-tricho-anal (MOTA) syndrome is caused by mutations in FREM1. Journal of Medical Genetics, 2011, 48, 375-382.	3.2	60
38	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
39	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
40	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
41	The DLX1and DLX2 genes and susceptibility to autism spectrum disorders. European Journal of Human Genetics, 2009, 17, 228-235.	2.8	75
42	History of genetics through philately - Carl Linnaeus (Carl von LinnÃ©). Clinical Genetics, 2008, 60, 104-106.	2.0	2
43	Unique disease heritage of the Dutchâ€German Mennonite population. American Journal of Medical Genetics, Part A, 2008, 146A, 1072-1087.	1.2	38
44	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
45	Genetic landmarks through philately - The Habsburg jaw. Clinical Genetics, 2008, 54, 283-284.	2.0	0
46	Fetal alcohol spectrum disorder: counting the invisible - mission impossible?. Archives of Disease in Childhood, 2008, 93, 721-722.	1.9	31
47	Development of Canadian screening tools for fetal alcohol spectrum disorder. Journal of Population Therapeutics and Clinical Pharmacology, 2008, 15, e344-66.	1.9	16
48	Manitoba oculotrichoanal (MOTA) syndrome: Report of eight new cases. American Journal of Medical Genetics, Part A, 2007, 143A, 853-857.	1.2	19
49	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
50	Autosomal recessive cerebellar hypoplasia in the Hutterite population. Developmental Medicine and Child Neurology, 2007, 47, 691-695.	2.1	1
51	Prevalence of Pervasive Developmental Disorders in Two Canadian Provinces. Journal of Policy and Practice in Intellectual Disabilities, 2006, 3, 164-172.	2.7	30
52	Autosomal recessive cerebellar hypoplasia in the Hutterite population. Developmental Medicine and Child Neurology, 2005, 47, 691.	2.1	34
53	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
54	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

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55	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
56	Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis. Cmaj, 2005, 172, S1-S21.	2.0	701
57	Identifying fetal alcohol spectrum disorder in primary care. Cmaj, 2005, 172, 628-630.	2.0	44
58	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
59	A notâ€soâ€â€œnewâ€•mental retardation syndrome. American Journal of Medical Genetics Part A, 2002, 111, 106-106.	2.4	6
60	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
61	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
62	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
63	Fetal hepatic haemangioendothelioma: a new association with elevated maternal serum alpha-fetoprotein. , 2000, 20, 432-434.		8
64	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
65	Genetic landmarks through philately - Henry Louis â€˜Louâ€™ Gehrig and amyotrophic lateral sclerosis. Clinical Genetics, 1999, 56, 425-427.	2.0	2
66	Agenesis of the penis: Patterns of associated malformations. , 1999, 84, 47-55.		49
67	Tibial agenesis, femoral duplication, and caudal midline anomalies. American Journal of Medical Genetics Part A, 1999, 85, 13-19.	2.4	25
68	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
69	Letters to the Editor. Journal of Developmental and Behavioral Pediatrics, 1999, 20, 72-73.	1.1	2
70	SAMS: Provisionally unique multiple congenital anomalies syndrome consisting of short stature, auditory canal atresia, mandibular hypoplasia, and skeletal abnormalities. , 1998, 75, 256-260.		7
71	Vertical transmission of the Ohdo blepharophimosis syndrome. American Journal of Medical Genetics Part A, 1998, 77, 144-148.	2.4	10
72	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

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73	Outcomes of Genetic Evaluation in Children with Pervasive Developmental Disorder. Journal of Developmental and Behavioral Pediatrics, 1998, 19, 321-325.	1.1	56
74	Bilateral sensorineural deafness and hydrocephalus due to foramen of Monro obstruction in sibs: A newly described autosomal recessive disorder. , 1997, 68, 350-356.		38
75	Proximal interstitial 6q deletion: A recognizable syndrome. American Journal of Medical Genetics Part A, 1997, 71, 353-356.	2.4	18
76	Evidence for Ritscherâ€Schinzel syndrome in Canadian native Indians. American Journal of Medical Genetics Part A, 1995, 56, 343-350.	2.4	33
77	Intragenic loss of function mutations demonstrate the primary role of FMR1 in fragile X syndrome. Nature Genetics, 1995, 10, 483-485.	21.4	152
78	Segregation analysis of rare autosomal folate sensitive fragile sites. American Journal of Medical Genetics Part A, 1993, 46, 165-171.	2.4	7
79	Mucinous cystadenoma of ovary in a patient with Williams syndrome. American Journal of Medical Genetics Part A, 1993, 46, 349-349.	2.4	12
80	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
81	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
82	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
83	Floating-Harbor syndrome and celiac disease. American Journal of Medical Genetics Part A, 1991, 38, 562-564.	2.4	22
84	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
85	Ulnar agenesis and endocardial fibroelastosis. American Journal of Medical Genetics Part A, 1990, 37, 258-260.	2.4	7
86	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
87	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
88	Familial duodenal atresia: A report of two families and review. American Journal of Medical Genetics Part A, 1989, 34, 442-444.	2.4	23
89	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
90	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

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91	Fragile X syndrome. Journal of Pediatrics, 1987, 110, 821-831.	1.8	116
92	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
93	Autism in fragile X females. American Journal of Medical Genetics Part A, 1986, 23, 375-380.	2.4	55
94	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
95	Zimmerman-Laband syndrome and profound mental retardation. American Journal of Medical Genetics Part A, 1986, 25, 543-547.	2.4	34
96	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
97	Successful pregnancy following continuous treatment with combination chemotherapy before conception and throughout pregnancy. Cancer, 1984, 54, 800-803.	4.1	69
98	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
99	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
100	Facial weakness and oligosyndactyly: ? independent variable features of familial type of the MÃ¶bius syndrome. Clinical Genetics, 1983, 24, 350-354.	2.0	18
101	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
102	The greig cephalopolysyndactyly syndrome in a canadian family. American Journal of Medical Genetics Part A, 1982, 13, 269-276.	2.4	18
103	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