

David D Weaver

List of Publications by Year in descending order

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57
papers

2,017
citations

304743

22
h-index

254184

43
g-index

57
all docs

57
docs citations

57
times ranked

2381
citing authors

#	ARTICLE	IF	CITATIONS
1	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
2	Occipital Horn Syndrome as a Result of Splice Site Mutations in ATP7A. No Activity of ATP7A Splice Variants Missing Exon 10 or Exon 15. Frontiers in Molecular Neuroscience, 2021, 14, 532291.	2.9	1
3	Response to Hamosh et al.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
4	Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. Clinical Dysmorphology, 2021, 30, 89-92.	0.3	2
5	Clinical spectrum of individuals with pathogenic <i>F1</i> missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
6	<i>EVENING</i> syndrome: A case report with novel variants in <i>HSPA9</i> and evidence of <i>HSPA9</i> gene dysfunction. American Journal of Medical Genetics, Part A, 2020, 182, 2501-2507.	1.2	6
7	Klinefelter's Syndrome with Maternal Uniparental Disomy X, Interstitial Xp22.31 Deletion, X-linked Ichthyosis, and Severe Central Nervous System Regression. Journal of Pediatric Genetics, 2020, 10, 222-229.	0.7	1
8	An Adolescent with a Rare <i>De Novo</i> Distal Trisomy 6p and Distal Monosomy 6q Chromosomal Combination. Case Reports in Genetics, 2020, 2020, 1-8.	0.2	1
9	An apparent new syndrome of extreme short stature, microcephaly, dysmorphic faces, intellectual disability, and a bone dysplasia of unknown etiology. American Journal of Medical Genetics, Part A, 2020, 182, 1562-1571.	1.2	0
10	PRC2 complex related dysfunction in overgrowth syndromes: A review of <i>EZH2</i> , <i>EED</i> , and <i>SUZ12</i> and their syndromic phenotypes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 519-531.	1.6	47
11	An unusual cause for Coffin-Lowry syndrome: Three brothers with a novel microduplication in <i>RPS6KA3</i> . American Journal of Medical Genetics, Part A, 2019, 179, 2357-2364.	1.2	1
12	The progression of Wiedemann-Steiner syndrome in adulthood and two novel variants in the <i>KMT2A</i> gene. American Journal of Medical Genetics, Part A, 2019, 179, 300-305.	1.2	12
13	Novel phenotype of 5p13.3-q11.2 duplication resulting from supernumerary marker chromosome 5: implications for management and genetic counseling. Molecular Cytogenetics, 2018, 11, 23.	0.9	1
14	Weaver Syndrome-Associated <i>EZH2</i> Protein Variants Show Impaired Histone Methyltransferase Function In Vitro. Human Mutation, 2016, 37, 301-307.	2.5	68
15	A distinct X-linked syndrome involving joint contractures, keloids, large optic cup-to-disc ratio, and renal stones results from a filamin A (<i>FLNA</i>) mutation. American Journal of Medical Genetics, Part A, 2016, 170, 881-890.	1.2	11
16	Setleis syndrome due to inheritance of the 1p36.22p36.21 duplication: evidence for lack of penetrance. Journal of Human Genetics, 2015, 60, 717-722.	2.3	12
17	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	6.2	47
18	Chromosome 1p36.22p36.21 duplications/triplication causes Setleis syndrome (focal facial dermal) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	1.2	15

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19	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogyriposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	6.2	171
20	Genitourinary Defects Associated with Genomic Deletions in 2p15 Encompassing OTX1. PLoS ONE, 2014, 9, e107028.	2.5	29
21	Prader-Willi syndrome: Are there population differences?. Clinical Genetics, 2008, 22, 292-294.	2.0	20
22	Distal 13q Deletion Syndrome and the VACTERL Association: Case report, literature review, and possible implications. American Journal of Medical Genetics Part A, 2001, 98, 137-144.	2.4	69
23	Partial urorectal septum malformation sequence: A report of 25 cases. American Journal of Medical Genetics Part A, 2001, 103, 99-105.	2.4	78
24	Exclusion of the branchio-oto-renal syndrome locus (EYA1) from patients with branchio-oculo-facial syndrome. , 2000, 91, 387-390.		19
25	Hypogonadism and CHARGE association. American Journal of Medical Genetics Part A, 2000, 94, 228-231.	2.4	34
26	Familial choanal atresia with maxillary hypoplasia, prognathism, and hypodontia. American Journal of Medical Genetics Part A, 2000, 95, 237-240.	2.4	6
27	Bone mineral density and laboratory evaluation of a type II autosomal dominant osteopetrosis carrier. , 1999, 85, 9-12.		7
28	Mild autosomal dominant hypophosphatasia: In utero presentation in two families. , 1999, 86, 410-415.		61
29	Lethal syndrome of skeletal dysplasia and progressive central nervous system degeneration. American Journal of Medical Genetics Part A, 1998, 77, 63-71.	2.4	8
30	Weaver syndrome: Autosomal dominant inheritance of the disorder. , 1998, 79, 305-310.		26
31	Molecular cytogenetic identification of four X chromosome duplications. , 1997, 68, 29-38.		28
32	Urorectal septum malformation sequence: Report of thirteen additional cases and review of the literature. American Journal of Medical Genetics Part A, 1997, 73, 456-462.	2.4	50
33	Urorectal septum malformation sequence: Report of thirteen additional cases and review of the literature. American Journal of Medical Genetics Part A, 1997, 73, 456-462.	2.4	3
34	Familial translocation resulting in Wolf-Hirschhorn syndrome in two related unbalanced individuals: Clinical evaluation of a 39-year-old man with Wolf-Hirschhorn syndrome. American Journal of Medical Genetics Part A, 1995, 55, 462-465.	2.4	27
35	Quality assurance in the clinical genetics setting?Report of a workshop. Journal of Genetic Counseling, 1994, 3, 169-198.	1.6	2
36	Branchio-oculo-facial syndrome: Broadening the spectrum. American Journal of Medical Genetics Part A, 1994, 49, 414-421.	2.4	22

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37	Molecular cytogenetic determination of a deletion/duplication of 1q that results in a trisomy 18 syndrome-like phenotype. American Journal of Medical Genetics Part A, 1994, 52, 178-183.	2.4	23
38	Asymmetric and symmetric long bone bowing in two sibs: An apparently new bone dysplasia. American Journal of Medical Genetics Part A, 1993, 47, 1072-1077.	2.4	7
39	Bone dysplasias: The prenatal diagnostic challenge. American Journal of Medical Genetics Part A, 1990, 36, 488-494.	2.4	20
40	Holoprosencephaly, ear abnormalities, congenital heart defect, and microphallus in a patient with 11q-mosaicism. American Journal of Medical Genetics Part A, 1989, 32, 178-181.	2.4	26
41	Familial distal arthrogyriposis with craniofacial abnormalities: A new subtype of type II?. American Journal of Medical Genetics Part A, 1989, 33, 231-237.	2.4	19
42	Neural tube defects and omphalocele in trisomy 18. Clinical Genetics, 1988, 34, 98-103.	2.0	18
43	A counseling guide to the Martin-Bell syndrome. American Journal of Medical Genetics Part A, 1987, 26, 39-44.	2.4	9
44	Presymptomatic testing for Huntington chorea: Guidelines for moral and social accountability. American Journal of Medical Genetics Part A, 1987, 26, 247-257.	2.4	28
45	Twenty-four ways to have children. American Journal of Medical Genetics Part A, 1987, 26, 737-740.	2.4	0
46	A syndrome of microcephaly, eye anomalies, short stature, and mental deficiency. American Journal of Medical Genetics Part A, 1987, 26, 825-831.	2.4	13
47	Subclavian artery supply disruption sequence: Hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. American Journal of Medical Genetics Part A, 1986, 23, 903-918.	2.4	547
48	Congenital contractural arachnodactyly Report of four additional families and review of literature. Clinical Genetics, 1985, 27, 570-581.	2.0	61
49	In utero brain destruction resulting in collapse of the fetal skull, microcephaly, scalp rugae, and neurologic impairment: The fetal brain disruption sequence. American Journal of Medical Genetics Part A, 1984, 17, 509-521.	2.4	63
50	Ethical considerations in medical genetics-the prenatal diagnosis of hemophilia B. American Journal of Medical Genetics Part A, 1984, 17, 773-781.	2.4	1
51	Endocrine and genetic characterization of cousins with male pseudohermaphroditism: evidence that the Lubs phenotype can result from a mutation that alters the structure of the androgen receptor. Clinical Genetics, 1984, 26, 363-370.	2.0	2
52	Dicentric chromosome 13 and centromere inactivation. Human Genetics, 1983, 63, 332-337.	3.8	28
53	Hirschsprung disease: Etiologic implications of unsuccessful prenatal diagnosis. American Journal of Medical Genetics Part A, 1983, 16, 163-167.	2.4	12
54	Pericentric X inversion in dizygotic twins who differ in X chromosome inactivation and menstrual cycle function. Human Genetics, 1982, 62, 210-213.	3.8	19

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55	Brief cytogenetic case report: A 4.5-year-old girl with deletion 4q syndrome " de novo, 46,XX, del(4)(pterâ†'q31). American Journal of Medical Genetics Part A, 1982, 12, 103-107.	2.4	15
56	Hyperplexia: an inherited disorder of the startle response. Clinical Genetics, 1982, 21, 388-396.	2.0	45
57	Cartilage-hair hypoplasia, defective T-cell function, and Diamond-Blackfan anemia in an Amish child. American Journal of Medical Genetics Part A, 1981, 8, 291-297.	2.4	25