# David D Weaver

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6373472/publications.pdf

Version: 2024-02-01

| 57 | 2,017 | 304743 | 254184 |
|---|---|---|---|
| papers | citations | 22 h-index | 43 g-index |

| 57 | 57 | 57 | 2381 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Subclavian artery supply disruption sequence: Hypothesis of a vascular etiology for Poland, Klippel-Feil, and MÃ¶bius anomalies. American Journal of Medical Genetics Part A, 1986, 23, 903-918. | 2.4 | 547 |
| 2 | 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 |
| 3 | Clinical spectrum of individuals with pathogenic  N F1  missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315. | 2.5 | 80 |
| 4 | Partial urorectal septum malformation sequence: A report of 25 cases. American Journal of Medical Genetics Part A, 2001, 103, 99-105. | 2.4 | 78 |
| 5 | A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15. | 6.2 | 71 |
| 6 | 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 |
| 7 | Weaver Syndromeâ€Associated EZH2 Protein Variants Show Impaired Histone Methyltransferase Function In Vitro. Human Mutation, 2016, 37, 301-307. | 2.5 | 68 |
| 8 | 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 |
| 9 | Mild autosomal dominant hypophosphatasia: In utero presentation in two families. , 1999, 86, 410-415. | | 61 |
| 10 | Congenital contractural arachnodactyly Report of four additional families and review of literature. Clinical Genetics, 1985, 27, 570-581. | 2.0 | 61 |
| 11 | 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 |
| 12 | Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531. | 6.2 | 47 |
| 13 | PRC2â€complex related dysfunction in overgrowth syndromes: A review of EZH2, EED, and SUZ12 and their syndromic phenotypes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 519-531. | 1.6 | 47 |
| 14 | Hyperexplexia: an inherited disorder of the startle response. Clinical Genetics, 1982, 21, 388-396. | 2.0 | 45 |
| 15 | Hypogonadism and CHARGE association. American Journal of Medical Genetics Part A, 2000, 94, 228-231. | 2.4 | 34 |
| 16 | Genitourinary Defects Associated with Genomic Deletions in 2p15 Encompassing OTX1. PLoS ONE, 2014, 9, e107028. | 2.5 | 29 |
| 17 | Dicentric chromosome 13 and centromere inactivation. Human Genetics, 1983, 63, 332-337. | 3.8 | 28 |
| 18 | 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 |

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|---|---|---|---|
| 19 | Molecular cytogenetic identification of four X chromosome duplications. , 1997, 68, 29-38. | | 28 |
| 20 | 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 |
| 21 | Holoprosencephaly, ear abnormalities, congenital heart defect, and microphallus in a patient with 11qâ€“mosaicism. American Journal of Medical Genetics Part A, 1989, 32, 178-181. | 2.4 | 26 |
| 22 | Weaver syndrome: Autosomal dominant inheritance of the disorder. , 1998, 79, 305-310. | | 26 |
| 23 | 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 |
| 24 | Molecular cytogenetic determination of a deletion/duplication of 1q that results in a trisomy 18 syndromeâ€like phenotype. American Journal of Medical Genetics Part A, 1994, 52, 178-183. | 2.4 | 23 |
| 25 | Branchio-oculo-facial syndrome: Broadening the spectrum. American Journal of Medical Genetics Part A, 1994, 49, 414-421. | 2.4 | 22 |
| 26 | Bone dysplasias: The prenatal diagnostic challenge. American Journal of Medical Genetics Part A, 1990, 36, 488-494. | 2.4 | 20 |
| 27 | Prader-Willi syndrome: Are there population differences?. Clinical Genetics, 2008, 22, 292-294. | 2.0 | 20 |
| 28 | 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 |
| 29 | Familial distal arthrogryposis with craniofacial abnormalities: A new subtype of type II?. American Journal of Medical Genetics Part A, 1989, 33, 231-237. | 2.4 | 19 |
| 30 | Exclusion of the branchio-oto-renal syndrome locus (EYA1) from patients with branchio-oculo-facial syndrome. , 2000, 91, 387-390. | | 19 |
| 31 | Neural tube defects and omphalocele in trisomy 18. Clinical Genetics, 1988, 34, 98-103. | 2.0 | 18 |
| 32 | Brief cytogenetic case report: A 4.5-year-old girl with deletion 4q syndrome â€" de novo, 46,XX, del(4)(pterâ†'q31:). American Journal of Medical Genetics Part A, 1982, 12, 103-107. | 2.4 | 15 |
| 33 | Chromosome 1p36.22p36.21 duplications/triplication causes Setleis syndrome (focal facial dermal) Tj ETQq1 1 0.784314 rgBT /Overl | 1.2 | 15 |
| 34 | 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 |
| 35 | Hirschsprung disease: Etiologic implications of unsuccessful prenatal diagnosis. American Journal of Medical Genetics Part A, 1983, 16, 163-167. | 2.4 | 12 |
| 36 | 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 |

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|---|---|---|---|
| 37 | The progression of Wiedemannâ€“Steiner syndrome in adulthood and two novel variants in the KMT2A gene. American Journal of Medical Genetics, Part A, 2019, 179, 300-305. | 1.2 | 12 |
| 38 | A distinct Xâ€linked syndrome involving joint contractures, keloids, large optic cupâ€toâ€disc ratio, and renal stones results from a filamin A (FLNA) mutation. American Journal of Medical Genetics, Part A, 2016, 170, 881-890. | 1.2 | 11 |
| 39 | A counseling guide to the Martin-Bell syndrome. American Journal of Medical Genetics Part A, 1987, 26, 39-44. | 2.4 | 9 |
| 40 | 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 |
| 41 | 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 |
| 42 | Bone mineral density and laboratory evaluation of a type II autosomal dominant osteopetrosis carrier. , 1999, 85, 9-12. | | 7 |
| 43 | Familial choanal atresia with maxillary hypoplasia, prognathism, and hypodontia. American Journal of Medical Genetics Part A, 2000, 95, 237-240. | 2.4 | 6 |
| 44 | <scp>EVENâ€PLUS</scp> syndrome: A case report with novel variants in <scp>HSPA9</scp> and evidence of <scp>HSPA9</scp> gene dysfunction. American Journal of Medical Genetics, Part A, 2020, 182, 2501-2507. | 1.2 | 6 |
| 45 | 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 |
| 46 | Quality assurance in the clinical genetics setting?Report of a workshop. Journal of Genetic Counseling, 1994, 3, 169-198. | 1.6 | 2 |
| 47 | Endocrine and genetic characterization of cousins with male pseudohermaphroditism: evidence that the Lubs phenoâ€type can result from a mutation that alters the structure of the androgen receptor. Clinical Genetics, 1984, 26, 363-370. | 2.0 | 2 |
| 48 | Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. Clinical Dysmorphology, 2021, 30, 89-92. | 0.3 | 2 |
| 49 | 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 |
| 50 | 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 |
| 51 | An unusual cause for Coffinâ€“Lowry syndrome: Three brothers with a novel microduplication in RPS6KA3. American Journal of Medical Genetics, Part A, 2019, 179, 2357-2364. | 1.2 | 1 |
| 52 | 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 |
| 53 | An Adolescent with a Rare De Novo Distal Trisomy 6p and Distal Monosomy 6q Chromosomal Combination. Case Reports in Genetics, 2020, 2020, 1-8. | 0.2 | 1 |
| 54 | 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 |

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|---|---|---|---|
| 55 | Twenty-four ways to have children. American Journal of Medical Genetics Part A, 1987, 26, 737-740. | 2.4 | 0 |
| 56 | 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 |
| 57 | Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810. | 6.2 | 0 |