Judith G Hall

List of Publications by Year in descending order

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238 papers

10,806 citations

41344 49 h-index 96 g-index

267 all docs

267 docs citations

times ranked

267

6764 citing authors

#	Article	IF	CITATIONS
1	The spectrum of brain malformations and disruptions in twins. American Journal of Medical Genetics, Part A, 2021, 185, 2690-2718.	1.2	13
2	Continuing contributions of older academics. American Journal of Medical Genetics, Part A, 2021, 185, 647-657.	1.2	2
3	<scp>The mystery of monozygotic twinning</scp> I: What can Amyoplasia tell us about monozygotic twinning and the possible role of <scp>twin–twin</scp> transfusion?. American Journal of Medical Genetics, Part A, 2021, 185, 1816-1821.	1.2	5
4	<scp>The mystery of monozygotic twinning II</scp> : What can monozygotic twinning tell us about Amyoplasia from a review of the various mechanisms and types of monozygotic twinning?. American Journal of Medical Genetics, Part A, 2021, 185, 1822-1835.	1.2	5
5	Deformations associated with arthrogryposis. American Journal of Medical Genetics, Part A, 2021, 185, 2676-2682.	1.2	5
6	Northwest Indigenous Art and the Inspiring Spirits. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 254-260.	1.6	1
7	The Clubfoot, Le Piedâ€Bot. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 160-162.	1.6	1
8	The contributions of careful clinical observations: A legacy. American Journal of Medical Genetics, Part A, 2021, 185, 3202-3207.	1.2	1
9	Examining the Vanishing Twin Hypothesis of Neural Tube Defects: Application of an Epigenetic Predictor for Monozygotic Twinning. Twin Research and Human Genetics, 2021, 24, 155-159.	0.6	1
10	Using the Term Amyoplasia Loosely Can Lead to Confusion. American Journal of Human Genetics, 2020, 107, 1186-1187.	6.2	1
11	Recurrent constellations of embryonic malformations reâ€conceptualized as an overlapping group of disorders with shared pathogenesis. American Journal of Medical Genetics, Part A, 2020, 182, 2646-2661.	1.2	28
12	50 Years Ago in T J P. Journal of Pediatrics, 2020, 217, 72.	1.8	1
13	Central nervous system involvement in arthrogryposis multiplex congenita: Overview of causes, diagnosis, and care. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 345-353.	1.6	13
14	Research platform for children with arthrogryposis multiplex congenita: Findings from the pilot registry. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 427-435.	1.6	10
15	The diagnostic workup in a patient with AMC: Overview of the clinical evaluation and paraclinical analyses with review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 337-344.	1.6	15
16	A standardized autopsy protocol for arthrogryposis (multiple congenital contractures). American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 474-478.	1.6	5
17	Gene ontology analysis of arthrogryposis (multiple congenital contractures). American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 310-326.	1.6	36
18	Fetal arthrogryposis: Challenges and perspectives for prenatal detection and management. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 327-336.	1.6	29

#	Article	IF	CITATIONS
19	International multidisciplinary collaboration toward an annotated definition of arthrogryposis multiplex congenita. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 288-299.	1.6	46
20	Fetal cervical hyperextension in arthrogryposis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 354-362.	1.6	3
21	Reader response: Disability in adults with arthrogryposis is severe, partly invisible, and varies by genotype. Neurology, 2019, 92, 635.2-635.	1.1	O
22	Classification of arthrogryposis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 300-303.	1.6	30
23	Collaborating to advance interdisciplinary care for individuals with arthrogryposis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 273-276.	1.6	4
24	Summary of the 3rd international symposium on arthrogryposis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 277-279.	1.6	7
25	Development of an online registry for adults with arthrogryposis multiplex congenita: A protocol paper. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 454-460.	1.6	4
26	Twins and Twinning. , 2019, , 387-414.		2
27	Aerodigestive and communicative behaviors in anencephalic and hydranencephalic infants. Birth Defects Research, 2018, 111, 41-52.	1.5	4
28	Reflections on an academic career. Molecular Genetics & Enomic Medicine, 2017, 5, 187-195.	1.2	2
29	Background to the 2nd International Symposium on Arthrogryposis. Journal of Pediatric Orthopaedics, 2017, 37, S2-S3.	1.2	9
30	Longâ€term functional and mobility outcomes for individuals with arthrogryposis multiplex congenita. American Journal of Medical Genetics, Part A, 2017, 173, 1270-1278.	1.2	34
31	The Clinic Is My Laboratory: Life as a Clinical Geneticist. Annual Review of Genomics and Human Genetics, 2017, 18, 1-29.	6.2	4
32	Genetics and Classifications. Journal of Pediatric Orthopaedics, 2017, 37, S4-S8.	1.2	25
33	Using the skills of academic elders. South African Medical Journal, 2016, 106, 9.	0.6	1
34	Arthrogryposis as a Syndrome: Gene Ontology Analysis. Molecular Syndromology, 2016, 7, 101-109.	0.8	49
35	The early history of Pallister–Hall syndrome—Buried treasure of a sort. Gene, 2016, 589, 100-103.	2.2	11
36	Novel pathogenic variants and genes for myopathies identified by whole exome sequencing. Molecular Genetics &	1.2	43

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37	Review of Xâ€linked syndromes with arthrogryposis or early contractures—aid to diagnosis and pathway identification. American Journal of Medical Genetics, Part A, 2015, 167, 931-973.	1.2	18
38	Judith G. Hall: a genetic journey. Genetics in Medicine, 2015, 17, 91-92.	2.4	0
39	Arthrogryposis. , 2015, , 96-114.		1
40	Epigenetics: What does it mean for paediatric practice?. Paediatrics and Child Health, 2014, 19, 27-30.	0.6	6
41	Pallister–Hall syndrome has gone the way of modern medical genetics. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 414-418.	1.6	19
42	Fetal akinesia deformation sequence: Expanding the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 2643-2648.	1.2	12
43	Amyoplasia involving only the upper limbs or only involving the lower limbs with review of the relevant differential diagnoses. American Journal of Medical Genetics, Part A, 2014, 164, 859-873.	1.2	19
44	Arthrogryposis (multiple congenital contractures): Diagnostic approach to etiology, classification, genetics, and general principles. European Journal of Medical Genetics, 2014, 57, 464-472.	1.3	211
45	Oligohydramnios sequence revisited in relationship to arthrogryposis, with distinctive skin changes. American Journal of Medical Genetics, Part A, 2014, 164, 2775-2792.	1.2	17
46	Amyoplasia revisited. American Journal of Medical Genetics, Part A, 2014, 164, 700-730.	1.2	96
47	Gender and Generational Influences on the Pediatric Workforce and Practice. Pediatrics, 2014, 133, 1112-1121.	2.1	33
48	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
49	Arthrogryposes (Multiple Congenital Contractures)., 2013,, 1-101.		11
50	Twins and Twinning., 2013,, 1-20.		2
51	A mutation in <i>TGFB3</i> associated with a syndrome of low muscle mass, growth retardation, distal arthrogryposis and clinical features overlapping with marfan and loeys–dietz syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2040-2046.	1.2	83
52	Uterine structural anomalies and arthrogryposisâ€"death of an urban legend. American Journal of Medical Genetics, Part A, 2013, 161, 82-88.	1.2	15
53	Failure to identify antenatal multiple congenital contractures and fetal akinesia – proposal of guidelines to improve diagnosis. Prenatal Diagnosis, 2013, 33, 61-74.	2.3	59
54	The smallest of the small. Gene, 2013, 528, 55-57.	2.2	6

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55	Elements of morphology: General terms for congenital anomalies. American Journal of Medical Genetics, Part A, 2013, 161, 2726-2733.	1.2	101
56	Trajectory of an Academic Career. JAMA Pediatrics, 2013, 167, 108.	6.2	6
57	Pretibial linear vertical creases or indentations (shin dimples) associated with arthrogryposis. American Journal of Medical Genetics, Part A, 2013, 161, 737-744.	1.2	8
58	The role of patient advocacy/parent support groups. South African Medical Journal, 2013, 103, 1020.	0.6	17
59	Special section. Syndromeâ€specific growth charts. American Journal of Medical Genetics, Part A, 2012, 158A, 2645-2646.	1.2	8
60	Arthrogryposis (multiple congenital contractures) associated with failed termination of pregnancy. American Journal of Medical Genetics, Part A, 2012, 158A, 2214-2220.	1.2	11
61	We are failing to identify disorders of fetal movement – why?. Prenatal Diagnosis, 2012, 32, 919-920.	2.3	10
62	Over the years, I hope I've learned a few things to pass along!. Paediatrics and Child Health, 2011, 16, 387-388.	0.6	0
63	Importance of Muscle Movement for Normal Craniofacial Development. Journal of Craniofacial Surgery, 2010, 21, 1336-1338.	0.7	19
64	Review and hypothesis: Syndromes with severe intrauterine growth restriction and very short statureâ€"Are they related to the epigenetic mechanism(s) of fetal survival involved in the developmental origins of adult health and disease?. American Journal of Medical Genetics, Part A, 2010, 152A, 512-527.	1.2	28
65	New palpebral fissure measurements. American Journal of Medical Genetics, Part A, 2010, 152A, 1870-1870.	1.2	6
66	Prevalence of multiple congenital contractures including arthrogryposis multiplex congenita in Alberta, Canada, and a strategy for classification and coding. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 1057-1061.	1.6	107
67	Victor A. McKusick, M.D.: A clinician's clinician. American Journal of Medical Genetics, Part A, 2009, 149A, 1105-1107.	1.2	2
68	Elements of morphology: Standard terminology for the nose and philtrum. American Journal of Medical Genetics, Part A, 2009, 149A, 61-76.	1.2	71
69	Penaâ€Shokeir phenotype (Fetal akinesia deformation sequence) revisited. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 677-694.	1.6	92
70	Ambulatory Activity in Youth With Arthrogryposis. Journal of Pediatric Orthopaedics, 2009, 29, 214-217.	1.2	40
71	Victor A. McKusick, M.D.: A legend in his own time. Clinical Genetics, 2008, 74, 293-295.	2.0	1
72	Localized acalvaria with craniosynostosis. Clinical Dysmorphology, 2008, 17, 165-168.	0.3	0

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73	Arthrogryposis Multiplex Congenita (Amyoplasia). Journal of Pediatric Orthopaedics, 2007, 27, 594-600.	1.2	135
74	Achondroplasia. Lancet, The, 2007, 370, 162-172.	13.7	456
75	Complex Inheritance Pattern Resembling Autosomal Recessive Inheritance Involving a Microdeletion in Thrombocytopenia–Absent Radius Syndrome. American Journal of Human Genetics, 2007, 80, 232-240.	6.2	290
76	The importance of the fetal origins of adult disease for geneticists. Clinical Genetics, 2007, 72, 67-73.	2.0	22
77	Preparing a manuscript for publication: A user-friendly guide. Paediatrics and Child Health, 2006, 11, 339-342.	0.6	7
78	Festschrift reflection. American Journal of Medical Genetics, Part A, 2006, 140A, 114-114.	1.2	0
79	Re: Majewski osteodysplastic primordial dwarfism type II (MOPD II) complicated by stroke: Clinical report and review of cerebral vascular anomalies [Brancati et al., 2005: Am J Med Genet 139A:212–215]. American Journal of Medical Genetics, Part A, 2006, 140A, 1356-1356.	1.2	4
80	Editorial independence for CMAJ: signposts along the road. Cmaj, 2006, 175, 453-453.	2.0	2
81	A meeting of minds: interdisciplinary research in the health sciences in Canada. Cmaj, 2006, 175, 763-771.	2.0	68
82	Re: Distal arthrogryposis in two sisters born to different fathers [Hwu et al. 2004. Am J Med Genet 125A:100-101.]. American Journal of Medical Genetics, Part A, 2005, 136A, 415-415.	1.2	5
83	Re: Microcephalic osteodysplastic primordial dwarfism with severe microdontia and skin anomalies [Kantaputra et al. 2004. Am J Med Genet 130A:181-190]. American Journal of Medical Genetics, Part A, 2005, 135A, 114-114.	1.2	3
84	A syndrome characterized by contractures and pterygia of upper body associated with umbilical hernia, short stature, and distinctive face in an Arabic family. American Journal of Medical Genetics, Part A, 2005, 138A, 236-240.	1.2	6
85	The Challenge of Developing Career Pathways for Senior Academic Pediatricians. Pediatric Research, 2005, 57, 914-919.	2.3	24
86	Health Supervision for Children With Achondroplasia. Pediatrics, 2005, 116, 771-783.	2.1	185
87	Introductory Speech for Robert J. Gorlin*. American Journal of Human Genetics, 2005, 76, 215.	6.2	O
88	Pediatricians beware: The age of ARTs is upon us. Journal of Pediatrics, 2005, 146, 450-452.	1.8	8
89	Epigenetics is Here to Stay. Journal of Pediatrics, 2005, 147, 427-428.	1.8	4
90	PRIMARY DISORDERS OF BONE AND CONNECTIVE TISSUES. , 2005, , 744-765.		O

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91	How is the progress in genetics relevant to children's health care. Paediatrics and Child Health, 2004, 9, 213-214.	0.6	2
92	Arthrogryposis multiplex and related congenital disorders. Handbook of Clinical Neurophysiology, 2004, , 387-401.	0.0	0
93	Long-term follow-up of three individuals with Kabuki syndrome. American Journal of Medical Genetics Part A, 2004, 125A, 191-200.	2.4	17
94	RE: Segmental neurofibromatosis in childhood [Listernick et al., 2003: Am. J. Med. Genet. 121A:132-135.]. American Journal of Medical Genetics Part A, 2004, 128A, 222-222.	2.4	0
95	Behavioral pattern profile: A tool for the description of behavior to be used in the genetics clinic. American Journal of Medical Genetics Part A, 2004, 128A, 389-395.	2.4	5
96	Majewski osteodysplastic primordial dwarfism type II (MOPD II): Natural history and clinical findings. American Journal of Medical Genetics Part A, 2004, 130A, 55-72.	2.4	132
97	Re: Down syndrome and folic acid deficiency. American Journal of Medical Genetics Part A, 2004, 131A, 327-327.	2.4	2
98	Tibial aplasia, lower extremity mirror image polydactyly, brachyphalangy, craniofacial dysmorphism and genital hypoplasia: further delineation and mutational analysis. Clinical Dysmorphology, 2004, 13, 63-69.	0.3	13
99	Morphogenesis: clinical natural history and imaging information on patients included in reports. Pediatric Radiology, 2003, 33, 146-146.	2.0	3
100	Poland anomaly?report of an unusual family. American Journal of Medical Genetics Part A, 2003, 118A, 180-183.	2.4	22
101	Morphogenesis: Re: Clinical, natural history, and imaging information on patients included in reports. American Journal of Medical Genetics Part A, 2003, 119A, 93-93.	2.4	5
102	A clinician's plea. Nature Genetics, 2003, 33, 440-442.	21.4	38
103	Twinning. Lancet, The, 2003, 362, 735-743.	13.7	583
104	So you think your mother is always looking over your shoulder?—She may be in your shoulder!. Journal of Pediatrics, 2003, 142, 233-234.	1.8	8
105	American Pediatric Society Presidential Address 2002: The Third Third. Pediatric Research, 2003, 53, 516-520.	2.3	4
106	Another adult with Meier-Gorlin syndrome ??? insights into the natural history. Clinical Dysmorphology, 2003, 12, 167-169.	0.3	2
107	Another adult with Meier-Gorlin syndrome - insights into the natural history. Clinical Dysmorphology, 2003, 12, 167-169.	0.3	17
108	Individualized medicine. What the genetic revolution will bring to health care in the 21st century. Canadian Family Physician, 2003, 49, 12-3, 15-7.	0.4	7

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109	Don't use the term ?amyoplasia? loosely. American Journal of Medical Genetics Part A, 2002, 111, 344-344.	2.4	5
110	Detection of Y-specific sequences in patients with Turner syndrome. American Journal of Medical Genetics Part A, 2002, 113, 114-114.	2.4	8
111	Clinical and radiologic information or photographs. Pediatric Radiology, 2002, 32, 609-609.	2.0	1
112	Paediatrician Resource Survey: Preliminary results suggest some urgency. Paediatrics and Child Health, 2001, 6, 12-13.	0.6	2
113	When is careless conception a form of child abuse? Lessons from maternal phenylketonuria. Journal of Pediatrics, 2000, 136, 12-13.	1.8	5
114	U-P- What?. Journal of Pediatrics, 1999, 134, 9-10.	1.8	2
115	See One, Do One, Teach One. Pediatrics, 1999, 103, 155-155.	2.1	8
116	Human Diseases and Genomic Imprinting. Results and Problems in Cell Differentiation, 1999, 25, 119-132.	0.7	7
117	Genetics of neural tube defects. Mental Retardation and Developmental Disabilities Research Reviews, 1998, 4, 269-281.	3.6	29
118	A bone is not a bone is not a bone. Journal of Pediatrics, 1998, 133, 5-6.	1.8	3
119	The Impact of Birth Defects and Genetic Diseases. JAMA Pediatrics, 1997, 151, 1082.	3.0	17
120	Arthrogryposis Multiplex Congenita. Journal of Pediatric Orthopaedics Part B, 1997, 6, 159-166.	0.6	274
121	Terathanasia, folic acid, and birth defects. Lancet, The, 1997, 350, 1322.	13.7	13
122	Neonatal personnel in Canada. Paediatrics and Child Health, 1997, 2, 193-197.	0.6	7
123	Give the embryo a chance. Nature Medicine, 1997, 3, 24-25.	30.7	1
124	Photographic documentation of syndrome diagnosis. , 1997, 68, 487-487.		3
125	Mosaicism in pseudoachondroplasia. American Journal of Medical Genetics Part A, 1997, 70, 287-291.	2.4	41
126	Twinning: mechanisms and genetic implications. Current Opinion in Genetics and Development, 1996, 6, 343-347.	3.3	99

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127	Twins and twinning. , 1996, 61, 202-204.		85
128	Arthrogryposis associated with unsuccessful attempts at termination of pregnancy., 1996, 63, 293-300.		35
129	Syndrome of multiple epiphyseal dysplasia (ribbing type) with rhizomelic shortness, cleft palate, and micrognathia in two unrelated patients. American Journal of Medical Genetics Part A, 1996, 63, 55-61.	2.4	3
130	Segregation analysis of microcephaly. , 1996, 65, 226-234.		6
131	Medial-Approach Open Reduction of Hip Dislocation in Amyoplasia-Type Arthrogryposis. Journal of Pediatric Orthopaedics, 1996, 16, 127-130.	1.2	18
132	Medial-Approach Open Reduction of Hip Dislocation in Amyoplasia-Type Arthrogryposis. Journal of Pediatric Orthopaedics, 1996, 16, 127-130.	1.2	49
133	Dominant distal arthrogryposis in a Maori family with marked variability of expression. American Journal of Medical Genetics Part A, 1995, 55, 414-419.	2.4	28
134	Recommendations for Diagnosis, Treatment, and Management of Individuals with Turner Syndrome. , 1994, 4, 351-358.		56
135	Evidence for multiâ€site closure of the neural tube in humans. American Journal of Medical Genetics Part A, 1993, 47, 723-743.	2.4	364
136	Genomic Imprinting and Its Clinical Implications. New England Journal of Medicine, 1992, 326, 827-829.	27.0	40
137	Nontraditional Inheritance. Pediatric Clinics of North America, 1992, 39, 335-348.	1.8	12
138	Fellowships and Career Development in Dysmorphology and Clinical Genetics. Pediatric Clinics of North America, 1992, 39, 349-362.	1.8	3
139	Fetal hypokinesia sequence caused by maternal autoimmune disorder?. American Journal of Medical Genetics Part A, 1992, 43, 1047-1048.	2.4	7
140	Genomic imprinting. Current Opinion in Genetics and Development, 1991, 1, 34-39.	3.3	19
141	The New Genetics and Its Relevance to Orthopedics. Clinical Orthopaedics and Related Research, 1991,	1.5	O
	&NA, 10???15.		
142	Neurofibromatosis I: Predicting the relation of gene structure to gene function. American Journal of Medical Genetics Part A, 1991, 38, 135-135.	2,4	9
142 143	Neurofibromatosis I: Predicting the relation of gene structure to gene function. American Journal of	2.4	9

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145	Congenital Anomalies: An Increasingly Important Cause of Mortality and Workload in a Neonatal intensive Care Unit. American Journal of Perinatology, 1991, 8, 164-169.	1.4	20
146	Turner Syndrome and Its Variants. Pediatric Clinics of North America, 1990, 37, 1421-1440.	1.8	139
147	Three-generation dominant transmission of the Silver-Russell syndrome. American Journal of Medical Genetics Part A, 1990, 35, 245-250.	2.4	89
148	Familial renal hypophosphatemia, minor facial anomalies, intracerebral calcifications, and non-rachitic bone changes: Apparently new syndrome?. American Journal of Medical Genetics Part A, 1990, 35, 406-414.	2.4	6
149	Could acrocallosal syndrome and Greig syndrome affect the same developmental gene?. American Journal of Medical Genetics Part A, 1990, 36, 368-368.	2.4	5
150	Partial expression of Angelman syndrome in mother most likely to be due to mosaicism involving both somatic and germline cells. American Journal of Medical Genetics Part A, 1990, 36, 369-369.	2.4	2
151	Apparent postnatal onset of some manifestations of the Wiedemann-Beckwith syndrome. American Journal of Medical Genetics Part A, 1990, 36, 434-439.	2.4	41
152	A sibship with Roberts/SC phocomelia syndrome. American Journal of Medical Genetics Part A, 1990, 37, 18-22.	2.4	12
153	Syndrome of mental retardation, facial anomalies, hypopituitarism, and distal arthrogryposis in sibs. American Journal of Medical Genetics Part A, 1990, 37, 65-70.	2.4	16
154	Congenital rubella syndrome associated with calcific epiphyseal stippling and peroxisomal dysfunction. Journal of Pediatrics, 1990, 116, 88-94.	1.8	26
155	Somatic and germâ€line mosaicism in autosomal dominant antecubital pterygium. Clinical Genetics, 1990, 37, 160-160.	2.0	2
156	How imprinting is relevant to human disease. Development (Cambridge), 1990, 108, 141-148.	2.5	18
157	Hydrocephalus in achondroplasia: the possible role of intracranial venous hypertension. Journal of Neurosurgery, 1989, 71, 42-48.	1.6	108
158	De novo reciprocal 1p;2q translocation in a child with multiple congenital anomalies/mental retardation syndrome. American Journal of Medical Genetics Part A, 1989, 32, 36-41.	2.4	4
159	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. American Journal of Medical Genetics Part A, 1989, 32, 93-99.	2.4	56
160	Congenital shortness of the costocoracoid ligament. American Journal of Medical Genetics Part A, 1989, 33, 444-446.	2.4	6
161	DETECTING MATERNAL CELL CONTAMINATION IN PRENATAL DIAGNOSIS. Lancet, The, 1989, 333, 1074-1075.	13.7	11
162	Perinatal and first year followâ€up of patients with Praderâ€Willi syndrome: normal size of hands and feet. Clinical Genetics, 1989, 35, 161-166.	2.0	16

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163	An unusual bandlike web in an infant with lethal multiple pterygium syndrome. American Journal of Medical Genetics Part A, 1988, 30, 763-769.	2.4	12
164	Bleeding diathesis in Noonan syndrome: A common association. American Journal of Medical Genetics Part A, 1988, 31, 305-317.	2.4	74
165	Congenital abnormalities in two sibs exposed to valproic acid in utero. American Journal of Medical Genetics Part A, 1988, 31, 369-373.	2.4	15
166	Juvenile galactosialidosis in a white male: A new variant. American Journal of Medical Genetics Part A, 1988, 31, 887-901.	2.4	17
167	Comments on "amyoplasia congenita-like condition and maternal malathion exposure― Is all amyoplasia amyoplasia?. Teratology, 1988, 38, 493-494.	1.6	5
168	Kyphosis in achondroplasia: Probably preventable. Journal of Pediatrics, 1988, 112, 166-167.	1.8	30
169	ABNORMALITIES OF CORPUS CALLOSUM IN PATIENTS WITH INHERITED METABOLIC DISEASES. Lancet, The, 1988, 332, 451.	13.7	32
170	Natural History of Human Chondrodysplasias. Pathology and Immunopathology Research, 1988, 7, 81-84.	0.8	0
171	Familial limb deficiency. Clinical Genetics, 1988, 34, 141-142.	2.0	8
172	Mild expression of the Pfeiffer syndrome. Clinical Genetics, 1988, 34, 144-144.	2.0	2
173	The Natural History of Achondroplasia. , 1988, 48, 3-9.		40
174	Lymphedema in Noonan syndrome: Clues to pathogenesis and prenatal diagnosis and review of the literature. American Journal of Medical Genetics Part A, 1987, 27, 841-856.	2.4	131
175	Gonadal mosaicism in pseudoachondroplasia. American Journal of Medical Genetics Part A, 1987, 28, 143-151.	2.4	67
176	Thanatophoric dysplasia and cloverleaf skull. American Journal of Medical Genetics Part A, 1987, 28, 167-179.	2.4	99
177	Familial breast cancer in males: A case report and review of the literature. Cancer, 1986, 58, 2736-2739.	4.1	39
178	Analysis of Pena Shokeir phenotype. American Journal of Medical Genetics Part A, 1986, 25, 99-117.	2.4	165
179	Studies of human achondroplasia: Oxidative metabolism in tissue culture cells. Teratology, 1986, 33, 9-13.	1.6	9
180	Growth curves for height in Noonan syndrome. Clinical Genetics, 1986, 30, 150-153.	2.0	99

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181	Prenatal Detection of Connective Tissue Disorders. , 1986, , 701-721.		O
182	Genetic Aspects of Arthrogryposis. Clinical Orthopaedics and Related Research, 1985, &NA, 44???53.	1.5	81
183	Familial multiple exostoses—no chromosome 8 deletion observed. American Journal of Medical Genetics Part A, 1985, 22, 639-640.	2.4	8
184	Medical Genetics. JAMA - Journal of the American Medical Association, 1985, 254, 2296.	7.4	1
185	Partial deletion of the short arm of chromosome 3 (3p25 â†' 3pter) Further delineation of the clinical phenotype. Clinical Genetics, 1985, 27, 402-407.	2.0	21
186	The lethal multiple pterygium syndromes. American Journal of Medical Genetics Part A, 1984, 17, 803-807.	2.4	111
187	Achondroplasia: Unexpected familial recurrence. American Journal of Medical Genetics Part A, 1984, 19, 245-250.	2.4	30
188	Vitamin A: A newly recognized human teratogen. Harbinger of things to come?. Journal of Pediatrics, 1984, 105, 583-584.	1.8	35
189	Apnea and sudden unexpected death in infants with achondroplasia. Journal of Pediatrics, 1984, 104, 342-348.	1.8	183
190	Prenatal diagnosis of genetic osteochondrodysplasias. American Journal of Medical Genetics Part A, 1983, 16, 285-287.	2.4	6
191	Association between age of onset and parental inheritance in Huntington chorea. American Journal of Medical Genetics Part A, 1983, 16, 289-290.	2.4	13
192	Neuropathologic findings in the spinal cords of 10 infants with arthrogryposis. Journal of the Neurological Sciences, 1983, 58, 89-102.	0.6	57
193	Limb pterygium syndromes: A review and report of eleven patients. American Journal of Medical Genetics Part A, 1982, 12, 377-409.	2.4	164
194	Head growth in achondroplasia: Use of ultrasound studies. American Journal of Medical Genetics Part A, 1982, 13, 105-105.	2.4	9
195	Kaufman syndrome. American Journal of Medical Genetics Part A, 1981, 8, 395-396.	2.4	5
196	Familial insertional translocation of a portion of 3q into 11q resulting in duplication and deletion of region 3q22.1â†'q24 in different offspring. American Journal of Medical Genetics Part A, 1981, 9, 105-111.	2.4	41
197	Comments on the Neu-Laxova syndrome and CAD complex. American Journal of Medical Genetics Part A, 1981, 9, 165-175.	2.4	45
198	Prometaphase chromosomes in five patients with the Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1981, 10, 179-186.	2.4	25

#	Article	IF	CITATIONS
199	Cerebroarthrodigital syndrome: A newly recognized formal genesis syndrome in three patients with apparent arthromyodysplasia and sacral agenesis, brain malformation and digital hypoplasia. American Journal of Medical Genetics Part A, 1980, 5, 13-24.	2.4	35
200	Genetic counseling for adoptees at risk for specific inherited disorders. American Journal of Medical Genetics Part A, 1980, 5, 157-164.	2.4	8
201	Endocardial fibroelastosis, neurologic dysfunction and unusual facial appearance in two brothers, coincidentally associated with dominantly inherited macrocephaly. American Journal of Medical Genetics Part A, 1980, 5, 271-276.	2.4	5
202	Autosomal recessive acrocephalosyndactyly revisited. American Journal of Medical Genetics Part A, 1980, 5, 423-424.	2.4	5
203	Autosomal-dominant inheritance of distal arthrogryposis. American Journal of Medical Genetics Part A, 1980, 6, 163-169.	2.4	17
204	Gardner syndrome and periampullary malignancy. American Journal of Medical Genetics Part A, 1980, 6, 205-219.	2.4	73
205	Congenital hypothalamic hamartoblastoma, hypopituitarism, imperforate anus, and postaxial polydactyly—a new syndrome? Part II: Neuropathological considerations. American Journal of Medical Genetics Part A, 1980, 7, 75-83.	2.4	92
206	Prenatal genetic diagnosis and elective abortion in women over 35: Utilization and relative impact on the birth prevalence of Down syndrome in Washington State. American Journal of Medical Genetics Part A, 1980, 7, 375-381.	2.4	23
207	Lower limb anomalies in the thrombocytopenia absent-radius (TAR) syndrome. American Journal of Medical Genetics Part A, 1980, 7, 523-528.	2.4	22
208	Rothmund-Thomson Syndrome With Severe Dwarfism. JAMA Pediatrics, 1980, 134, 165.	3.0	9
209	Prenatal Diagnosis of Congenital Bullous Ichthyosiform Erythroderma (Epidermolytic) Tj ETQq1 1 0.784314 rgBT /	Overlock 27.0	19 ₀ f 50 34
210	Risks of anticoagulation during pregnancy. American Heart Journal, 1980, 100, 761-762.	2.7	15
211	Maternal and fetal sequelae of anticoagulation during pregnancy. American Journal of Medicine, 1980, 68, 122-140.	1.5	975
212	X-Linked Cutis Laxa. New England Journal of Medicine, 1980, 303, 61-65.	27.0	205
213	The summitt syndrome: Observations on a third case. American Journal of Medical Genetics Part A, 1979, 3, 27-33.	2.4	5
214	Frequency and characteristics of birth defects admissions to a pediatric hospital in Venezuela. American Journal of Medical Genetics Part A, 1979, 3, 359-369.	2.4	17
215	Failure of early prenatal diagnosis in classic achondroplasia. American Journal of Medical Genetics Part A, 1979, 3, 371-375.	2.4	12
216	Fibrodysplasia ossificans progressiva (myositis ossificans progressiva) treatment with disodium etidronate. Journal of Pediatrics, 1979, 94, 679-680.	1.8	8

#	Article	IF	CITATIONS
217	An autosomal dominantly inherited syndrome of facial asymmetry, esotropia, amblyopia, and submucous cleft palate (Bencze syndrome). Clinical Genetics, 1979, 16, 301-304.	2.0	6
218	No evidence for chromosomal mosaicism in multiple tissues of 10 patients with 45 XO Turner syndrome. Clinical Genetics, 1979, 15, 22-28.	2.0	9
219	Isolated congenital ectopia lentis with autosomal dominant inheritance. Clinical Genetics, 1979, 15, 97-109.	2.0	16
220	A new variety of spondyloepiphyseal dysplasia characterized by punctate corneal dystrophy and abnormal dermal collagen fibrils. Human Genetics, 1978, 40, 157-169.	3.8	28
221	The frequency and financial burden of genetic disease in a pediatric hospital. American Journal of Medical Genetics Part A, 1978, 1, 417-436.	2.4	102
222	The phenotypic variability of diastrophic dysplasia. Journal of Pediatrics, 1978, 93, 609-613.	1.8	67
223	Standard growth curves for achondroplasia. Journal of Pediatrics, 1978, 93, 435-438.	1.8	266
224	Children of Incest: When To Suspect and How To Evaluate?. JAMA Pediatrics, 1978, 132, 1045.	3.0	2
225	Microphallus, Growth Hormone Deficiency, and Hypoglycemia in Russell-Silver Syndrome. JAMA Pediatrics, 1978, 132, 1149.	3.0	8
226	Additional information on familial essential (benign) chorea. Clinical Genetics, 1978, 14, 271-272.	2.0	6
227	The 2p Partial Trisomy Syndrome. JAMA Pediatrics, 1977, 131, 1405.	3.0	1
228	Multiple congenital anomalies associated with oral anticoagulants. American Journal of Obstetrics and Gynecology, 1977, 127, 191-198.	1.3	135
229	A lethal neonatal dwarfing condition with short ribs, polysyndactyly, cranial synostosis, cleft palate cardiovascular and urogenital anomalies and severe ossification defect. Teratology, 1977, 16, 345-350.	1.6	22
230	Acromesomelic dwarfism: Manifestations in childhood. American Journal of Medical Genetics Part A, 1977, 1, 87-100.	2.4	32
231	Warfarin and Fetal Abnormality: Reply. Lancet, The, 1976, 307, 1127-1127.	13.7	18
232	Chromosome 7 short arm deletion and craniosynostosis a 7p-syndrome. Human Genetics, 1976, 35, 117-123.	3.8	45
233	Dominantly inherited ptosis, strabismus and ectopic pupils. Clinical Genetics, 1976, 10, 21-26.	2.0	7
234	A New Arthrogryposis Syndrome With Facial and Limb Anomalies. JAMA Pediatrics, 1975, 129, 120.	3.0	3

#	Article	IF	CITATIONS
235	The Jeune syndrome (asphyxiating thoracic dystrophy) in an adult. American Journal of Medicine, 1975, 59, 857-862.	1.5	39
236	A pattern of craniofacial and limb defects secondary to aberrant tissue bands. Journal of Pediatrics, 1974, 84, 90-95.	1.8	114
237	DIASTROPHIC DWARFISM. Medicine (United States), 1972, 51, 41-59.	1.0	92
238	THROMBOCYTOPENIA WITH ABSENT RADIUS (TAR). Medicine (United States), 1969, 48, 411-440.	1.0	279