Judith G Hall

List of Publications by Year
in descending order

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238 papers	10,806 citations	41344 49 h-index	37204 96 g-index
267 all docs	267 docs citations	267 times ranked	6764 citing authors

#	Article	IF	Citations
1	Maternal and fetal sequelae of anticoagulation during pregnancy. American Journal of Medicine, 1980, 68, 122-140.	1.5	975
2	Twinning. Lancet, The, 2003, 362, 735-743.	13.7	583
3	Achondroplasia. Lancet, The, 2007, 370, 162-172.	13.7	456
4	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
5	Complex Inheritance Pattern Resembling Autosomal Recessive Inheritance Involving a Microdeletion in Thrombocytopeniaâ€“Absent Radius Syndrome. American Journal of Human Genetics, 2007, 80, 232-240.	6.2	290
6	THROMBOCYTOPENIA WITH ABSENT RADIUS (TAR). Medicine (United States), 1969, 48, 411-440.	1.0	279
7	Arthrogryposis Multiplex Congenita. Journal of Pediatric Orthopaedics Part B, 1997, 6, 159-166.	0.6	274
8	Standard growth curves for achondroplasia. Journal of Pediatrics, 1978, 93, 435-438.	1.8	266
9	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
10	X-Linked Cutis Laxa. New England Journal of Medicine, 1980, 303, 61-65.	27.0	205
11	Health Supervision for Children With Achondroplasia. Pediatrics, 2005, 116, 771-783.	2.1	185
12	Apnea and sudden unexpected death in infants with achondroplasia. Journal of Pediatrics, 1984, 104, 342-348.	1.8	183
13	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
14	Analysis of Pena Shokeir phenotype. American Journal of Medical Genetics Part A, 1986, 25, 99-117.	2.4	165
15	Limb pterygium syndromes: A review and report of eleven patients. American Journal of Medical Genetics Part A, 1982, 12, 377-409.	2.4	164
16	Turner Syndrome and Its Variants. Pediatric Clinics of North America, 1990, 37, 1421-1440.	1.8	139
17	Multiple congenital anomalies associated with oral anticoagulants. American Journal of Obstetrics and Gynecology, 1977, 127, 191-198.	1.3	135
18	Arthrogryposis Multiplex Congenita (Amyoplasia). Journal of Pediatric Orthopaedics, 2007, 27, 594-600.	1.2	135

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19	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
20	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
21	A pattern of craniofacial and limb defects secondary to aberrant tissue bands. Journal of Pediatrics, 1974, 84, 90-95.	1.8	114
22	The lethal multiple pterygium syndromes. American Journal of Medical Genetics Part A, 1984, 17, 803-807.	2.4	111
23	Hydrocephalus in achondroplasia: the possible role of intracranial venous hypertension. Journal of Neurosurgery, 1989, 71, 42-48.	1.6	108
24	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
25	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
26	Prenatal Diagnosis of Congenital Bullous Ichthyosiform Erythroderma (Epidermolytic) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 462 Td (Hyp	27.0	101
27	Elements of morphology: General terms for congenital anomalies. American Journal of Medical Genetics, Part A, 2013, 161, 2726-2733.	1.2	101
28	Thanatophoric dysplasia and cloverleaf skull. American Journal of Medical Genetics Part A, 1987, 28, 167-179.	2.4	99
29	Twinning: mechanisms and genetic implications. Current Opinion in Genetics and Development, 1996, 6, 343-347.	3.3	99
30	Growth curves for height in Noonan syndrome. Clinical Genetics, 1986, 30, 150-153.	2.0	99
31	Amyoplasia revisited. American Journal of Medical Genetics, Part A, 2014, 164, 700-730.	1.2	96
32	DIASTROPHIC DWARFISM. Medicine (United States), 1972, 51, 41-59.	1.0	92
33	Congenital hypothalamic hamartoblastoma, hypopituitarism, imperforate anus, and postaxial polydactylyâ€”a new syndrome? Part II: Neuropathological considerations. American Journal of Medical Genetics Part A, 1980, 7, 75-83.	2.4	92
34	Penaâ€Shokeir phenotype (Fetal akinesia deformation sequence) revisited. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 677-694.	1.6	92
35	Three-generation dominant transmission of the Silver-Russell syndrome. American Journal of Medical Genetics Part A, 1990, 35, 245-250.	2.4	89
36	Twins and twinning. , 1996, 61, 202-204.		85

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37	A mutation in TGFB3 associated with a syndrome of low muscle mass, growth retardation, distal arthrogryposis and clinical features overlapping with marfan and loeysâ€“dietz syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2040-2046.	1.2	83
38	Genetic Aspects of Arthrogryposis. Clinical Orthopaedics and Related Research, 1985, &NA;, 44???53.	1.5	81
39	Bleeding diathesis in Noonan syndrome: A common association. American Journal of Medical Genetics Part A, 1988, 31, 305-317.	2.4	74
40	Gardner syndrome and periampullary malignancy. American Journal of Medical Genetics Part A, 1980, 6, 205-219.	2.4	73
41	Elements of morphology: Standard terminology for the nose and philtrum. American Journal of Medical Genetics, Part A, 2009, 149A, 61-76.	1.2	71
42	A meeting of minds: interdisciplinary research in the health sciences in Canada. Cmaj, 2006, 175, 763-771.	2.0	68
43	The phenotypic variability of diastrophic dysplasia. Journal of Pediatrics, 1978, 93, 609-613.	1.8	67
44	Gonadal mosaicism in pseudoachondroplasia. American Journal of Medical Genetics Part A, 1987, 28, 143-151.	2.4	67
45	Failure to identify antenatal multiple congenital contractures and fetal akinesia â€“ proposal of guidelines to improve diagnosis. Prenatal Diagnosis, 2013, 33, 61-74.	2.3	59
46	Neuropathologic findings in the spinal cords of 10 infants with arthrogryposis. Journal of the Neurological Sciences, 1983, 58, 89-102.	0.6	57
47	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. American Journal of Medical Genetics Part A, 1989, 32, 93-99.	2.4	56
48	Recommendations for Diagnosis, Treatment, and Management of Individuals with Turner Syndrome. , 1994, 4, 351-358.		56
49	Deletion of chromosome 21 and normal intelligence: molecular definition of the lesion. Human Genetics, 1991, 87, 112-118.	3.8	49
50	Arthrogryposis as a Syndrome: Gene Ontology Analysis. Molecular Syndromology, 2016, 7, 101-109.	0.8	49
51	Medial-Approach Open Reduction of Hip Dislocation in Amyoplasia-Type Arthrogryposis. Journal of Pediatric Orthopaedics, 1996, 16, 127-130.	1.2	49
52	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
53	Chromosome 7 short arm deletion and craniosynostosis a 7p-syndrome. Human Genetics, 1976, 35, 117-123.	3.8	45
54	Comments on the Neu-Laxova syndrome and CAD complex. American Journal of Medical Genetics Part A, 1981, 9, 165-175.	2.4	45

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55	Novel pathogenic variants and genes for myopathies identified by whole exome sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 283-301.	1.2	43
56	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
57	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
58	Mosaicism in pseudoachondroplasia. American Journal of Medical Genetics Part A, 1997, 70, 287-291.	2.4	41
59	Genomic Imprinting and Its Clinical Implications. New England Journal of Medicine, 1992, 326, 827-829.	27.0	40
60	Ambulatory Activity in Youth With Arthrogryposis. Journal of Pediatric Orthopaedics, 2009, 29, 214-217.	1.2	40
61	The Natural History of Achondroplasia. , 1988, 48, 3-9.		40
62	The Jeune syndrome (asphyxiating thoracic dystrophy) in an adult. American Journal of Medicine, 1975, 59, 857-862.	1.5	39
63	Familial breast cancer in males:A case report and review of the literature. Cancer, 1986, 58, 2736-2739.	4.1	39
64	A clinician's plea. Nature Genetics, 2003, 33, 440-442.	21.4	38
65	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
66	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
67	Vitamin A: A newly recognized human teratogen. Harbinger of things to come?. Journal of Pediatrics, 1984, 105, 583-584.	1.8	35
68	Arthrogryposis associated with unsuccessful attempts at termination of pregnancy. , 1996, 63, 293-300.		35
69	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
70	Gender and Generational Influences on the Pediatric Workforce and Practice. Pediatrics, 2014, 133, 1112-1121.	2.1	33
71	Acromesomelic dwarfism: Manifestations in childhood. American Journal of Medical Genetics Part A, 1977, 1, 87-100.	2.4	32
72	ABNORMALITIES OF CORPUS CALLOSUM IN PATIENTS WITH INHERITED METABOLIC DISEASES. Lancet, The, 1988, 332, 451.	13.7	32

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73	Achondroplasia: Unexpected familial recurrence. American Journal of Medical Genetics Part A, 1984, 19, 245-250.	2.4	30
74	Kyphosis in achondroplasia: Probably preventable. Journal of Pediatrics, 1988, 112, 166-167.	1.8	30
75	Classification of arthrogryposis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 300-303.	1.6	30
76	Genetics of neural tube defects. Mental Retardation and Developmental Disabilities Research Reviews, 1998, 4, 269-281.	3.6	29
77	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
78	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
79	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
80	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
81	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
82	Congenital rubella syndrome associated with calcific epiphyseal stippling and peroxisomal dysfunction. Journal of Pediatrics, 1990, 116, 88-94.	1.8	26
83	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
84	Genetics and Classifications. Journal of Pediatric Orthopaedics, 2017, 37, S4-S8.	1.2	25
85	The Challenge of Developing Career Pathways for Senior Academic Pediatricians. Pediatric Research, 2005, 57, 914-919.	2.3	24
86	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
87	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
88	Lower limb anomalies in the thrombocytopenia absent-radius (TAR) syndrome. American Journal of Medical Genetics Part A, 1980, 7, 523-528.	2.4	22
89	Poland anomaly?report of an unusual family. American Journal of Medical Genetics Part A, 2003, 118A, 180-183.	2.4	22
90	The importance of the fetal origins of adult disease for geneticists. Clinical Genetics, 2007, 72, 67-73.	2.0	22

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91	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
92	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
93	Genomic imprinting. Current Opinion in Genetics and Development, 1991, 1, 34-39.	3.3	19
94	Importance of Muscle Movement for Normal Craniofacial Development. Journal of Craniofacial Surgery, 2010, 21, 1336-1338.	0.7	19
95	Pallisterâ€"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
96	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
97	Warfarin and Fetal Abnormality: Reply. Lancet, The, 1976, 307, 1127-1127.	13.7	18
98	Review of Xâ€linked syndromes with arthrogryposis or early contracturesâ€"aid to diagnosis and pathway identification. American Journal of Medical Genetics, Part A, 2015, 167, 931-973.	1.2	18
99	Medial-Approach Open Reduction of Hip Dislocation in Amyoplasia-Type Arthrogryposis. Journal of Pediatric Orthopaedics, 1996, 16, 127-130.	1.2	18
100	How imprinting is relevant to human disease. Development (Cambridge), 1990, 108, 141-148.	2.5	18
101	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
102	Autosomal-dominant inheritance of distal arthrogryposis. American Journal of Medical Genetics Part A, 1980, 6, 163-169.	2.4	17
103	Juvenile galactosialidosis in a white male: A new variant. American Journal of Medical Genetics Part A, 1988, 31, 887-901.	2.4	17
104	The Impact of Birth Defects and Genetic Diseases. JAMA Pediatrics, 1997, 151, 1082.	3.0	17
105	Another adult with Meier-Gorlin syndrome - insights into the natural history. Clinical Dysmorphology, 2003, 12, 167-169.	0.3	17
106	Long-term follow-up of three individuals with Kabuki syndrome. American Journal of Medical Genetics Part A, 2004, 125A, 191-200.	2.4	17
107	The role of patient advocacy/parent support groups. South African Medical Journal, 2013, 103, 1020.	0.6	17
108	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

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109	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
110	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
111	Isolated congenital ectopia lentis with autosomal dominant inheritance. Clinical Genetics, 1979, 15, 97-109.	2.0	16
112	Risks of anticoagulation during pregnancy. American Heart Journal, 1980, 100, 761-762.	2.7	15
113	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
114	Uterine structural anomalies and arthrogryposisâ€”death of an urban legend. American Journal of Medical Genetics, Part A, 2013, 161, 82-88.	1.2	15
115	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
116	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
117	Terathanasia, folic acid, and birth defects. Lancet, The, 1997, 350, 1322.	13.7	13
118	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
119	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
120	The spectrum of brain malformations and disruptions in twins. American Journal of Medical Genetics, Part A, 2021, 185, 2690-2718.	1.2	13
121	Failure of early prenatal diagnosis in classic achondroplasia. American Journal of Medical Genetics Part A, 1979, 3, 371-375.	2.4	12
122	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
123	A sibship with Roberts/SC phocomelia syndrome. American Journal of Medical Genetics Part A, 1990, 37, 18-22.	2.4	12
124	Nontraditional Inheritance. Pediatric Clinics of North America, 1992, 39, 335-348.	1.8	12
125	Fetal akinesia deformation sequence: Expanding the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 2643-2648.	1.2	12
126	DETECTING MATERNAL CELL CONTAMINATION IN PRENATAL DIAGNOSIS. Lancet, The, 1989, 333, 1074-1075.	13.7	11

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127	Arthrogryposis (multiple congenital contractures) associated with failed termination of pregnancy. American Journal of Medical Genetics, Part A, 2012, 158A, 2214-2220.	1.2	11
128	Arthrogryposes (Multiple Congenital Contractures). , 2013, , 1-101.		11
129	The early history of Pallisterâ€“Hall syndromeâ€”Buried treasure of a sort. Gene, 2016, 589, 100-103.	2.2	11
130	We are failing to identify disorders of fetal movement â€“ why?. Prenatal Diagnosis, 2012, 32, 919-920.	2.3	10
131	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
132	Rothmund-Thomson Syndrome With Severe Dwarfism. JAMA Pediatrics, 1980, 134, 165.	3.0	9
133	Head growth in achondroplasia: Use of ultrasound studies. American Journal of Medical Genetics Part A, 1982, 13, 105-105.	2.4	9
134	Studies of human achondroplasia: Oxidative metabolism in tissue culture cells. Teratology, 1986, 33, 9-13.	1.6	9
135	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
136	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
137	Background to the 2nd International Symposium on Arthrogryposis. Journal of Pediatric Orthopaedics, 2017, 37, S2-S3.	1.2	9
138	Microphallus, Growth Hormone Deficiency, and Hypoglycemia in Russell-Silver Syndrome. JAMA Pediatrics, 1978, 132, 1149.	3.0	8
139	Fibrodysplasia ossificans progressiva (myositis ossificans progressiva) treatment with disodium etidronate. Journal of Pediatrics, 1979, 94, 679-680.	1.8	8
140	Genetic counseling for adoptees at risk for specific inherited disorders. American Journal of Medical Genetics Part A, 1980, 5, 157-164.	2.4	8
141	Familial multiple exostosesâ€”no chromosome 8 deletion observed. American Journal of Medical Genetics Part A, 1985, 22, 639-640.	2.4	8
142	See One, Do One, Teach One. Pediatrics, 1999, 103, 155-155.	2.1	8
143	Detection of Y-specific sequences in patients with Turner syndrome. American Journal of Medical Genetics Part A, 2002, 113, 114-114.	2.4	8
144	So you think your mother is always looking over your shoulder?â€”She may be in your shoulder!. Journal of Pediatrics, 2003, 142, 233-234.	1.8	8

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145	Pediatricians beware: The age of ARTs is upon us. Journal of Pediatrics, 2005, 146, 450-452.	1.8	8
146	Familial limb deficiency. Clinical Genetics, 1988, 34, 141-142.	2.0	8
147	Special section. Syndromeâ€specific growth charts. American Journal of Medical Genetics, Part A, 2012, 158A, 2645-2646.	1.2	8
148	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
149	Fetal hypokinesia sequence caused by maternal autoimmune disorder?. American Journal of Medical Genetics Part A, 1992, 43, 1047-1048.	2.4	7
150	Neonatal personnel in Canada. Paediatrics and Child Health, 1997, 2, 193-197.	0.6	7
151	Preparing a manuscript for publication: A user-friendly guide. Paediatrics and Child Health, 2006, 11, 339-342.	0.6	7
152	Dominantly inherited ptosis, strabismus and ectopic pupils. Clinical Genetics, 1976, 10, 21-26.	2.0	7
153	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
154	Human Diseases and Genomic Imprinting. Results and Problems in Cell Differentiation, 1999, 25, 119-132.	0.7	7
155	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
156	Prenatal diagnosis of genetic osteochondrodysplasias. American Journal of Medical Genetics Part A, 1983, 16, 285-287.	2.4	6
157	Congenital shortness of the costocoracoid ligament. American Journal of Medical Genetics Part A, 1989, 33, 444-446.	2.4	6
158	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
159	Segregation analysis of microcephaly. , 1996, 65, 226-234.		6
160	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
161	Additional information on familial essential (benign) chorea. Clinical Genetics, 1978, 14, 271-272.	2.0	6
162	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

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163	New palpebral fissure measurements. American Journal of Medical Genetics, Part A, 2010, 152A, 1870-1870.	1.2	6
164	The smallest of the small. Gene, 2013, 528, 55-57.	2.2	6
165	Trajectory of an Academic Career. JAMA Pediatrics, 2013, 167, 108.	6.2	6
166	Epigenetics: What does it mean for paediatric practice?. Paediatrics and Child Health, 2014, 19, 27-30.	0.6	6
167	The summitt syndrome: Observations on a third case. American Journal of Medical Genetics Part A, 1979, 3, 27-33.	2.4	5
168	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
169	Autosomal recessive acrocephalosyndactyly revisited. American Journal of Medical Genetics Part A, 1980, 5, 423-424.	2.4	5
170	Kaufman syndrome. American Journal of Medical Genetics Part A, 1981, 8, 395-396.	2.4	5
171	Comments on â€œamyoplasia congenita-like condition and maternal malathion exposureâ€• Is all amyoplasia amyoplasia?. Teratology, 1988, 38, 493-494.	1.6	5
172	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
173	Syndrome of mental retardation and distal arthrogryposis in sibs. American Journal of Medical Genetics Part A, 1991, 41, 49-51.	2.4	5
174	When is careless conception a form of child abuse? Lessons from maternal phenylketonuria. Journal of Pediatrics, 2000, 136, 12-13.	1.8	5
175	Don't use the term ?amyoplasia? loosely. American Journal of Medical Genetics Part A, 2002, 111, 344-344.	2.4	5
176	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
177	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
178	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
179	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
180	<scp>The mystery of monozygotic twinning</scp> I: What can Amyoplasia tell us about monozygotic twinning and the possible role of <scp>twinâ€“twin</scp> transfusion?. American Journal of Medical Genetics, Part A, 2021, 185, 1816-1821.	1.2	5

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181	<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
182	Deformations associated with arthrogryposis. American Journal of Medical Genetics, Part A, 2021, 185, 2676-2682.	1.2	5
183	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
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