John M Opitz

List of Publications by Year
in descending order

221 papers	10,871 citations	50273 46 h-index	39667 94 g-index
238 all docs	238 docs citations	238 times ranked	9516 citing authors

#	Article	IF	Citations
1	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025.	28.9	1,725
2	A newly recognized syndromeof multiple congenital anomalies. Journal of Pediatrics, 1964, 64, 210-217.	1.8	657
3	Resynthesizing Evolutionary and Developmental Biology. Developmental Biology, 1996, 173, 357-372.	2.0	610
4	Noonan syndrome: A review. American Journal of Medical Genetics Part A, 1985, 21, 493-506.	2.4	425
5	Evaluation of mental retardation: Recommendations of a consensus conference. , 1997, 72, 468-477.		344
6	Opitz G/BBB syndrome, a defect of midline development, is due to mutations in a new RING finger gene on Xp22. Nature Genetics, 1997, 17, 285-291.	21.4	331
7	CNS anomalies and the midline as a â€œdevelopmental fieldâ€• American Journal of Medical Genetics Part A, 1982, 12, 443-455.	2.4	255
8	Mutations in mitochondrial histidyl tRNA synthetase HARS2 cause ovarian dysgenesis and sensorineural hearing loss of Perrault syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 6543-6548.	7.1	225
9	Germline mutations in DIS3L2 cause the Perlman syndrome of overgrowth and Wilms tumor susceptibility. Nature Genetics, 2012, 44, 277-284.	21.4	219
10	Smithâ€Lemliâ€Opitz syndromeâ€type II: Multiple congenital anomalies with male pseudohermaphroditism and frequent early lethality. American Journal of Medical Genetics Part A, 1987, 26, 45-57.	2.4	218
11	The DiGeorge anomaly as a developmental field defect. American Journal of Medical Genetics Part A, 1986, 25, 113-127.	2.4	183
12	A recurrent mutation in MED12 leading to R961W causes Opitz-Kaveggia syndrome. Nature Genetics, 2007, 39, 451-453.	21.4	179
13	The Brachmann-de Lange syndrome. American Journal of Medical Genetics Part A, 1985, 22, 89-102.	2.4	155
14	Isochromosome 12p mosaicism (Pallister mosaic aneuploidy or Pallisterâ€Killian syndrome): Report of 11 cases. American Journal of Medical Genetics Part A, 1987, 27, 257-274.	2.4	135
15	Studies of malformation syndromes of man XXXIII: The FG syndrome. An X-linked recessive syndrome of multiple congenital anomalies and mental retardation. European Journal of Pediatrics, 1974, 117, 1-18.	2.7	134
16	Fibular A/hypoplasia: Review and documentation of the fibular developmental field. American Journal of Medical Genetics Part A, 1986, 25, 215-238.	2.4	129
17	Rett syndrome at an institution for the developmentally disabled. American Journal of Medical Genetics Part A, 1986, 25, 85-97.	2.4	128
18	Errors of morphogenesis and developmental field theory. , 1998, 76, 291-296.		128

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19	The developmental field concept. American Journal of Medical Genetics Part A, 1985, 21, 1-11.	2.4	123
20	The syndromes of Sotos and Weaver: Reports and review. American Journal of Medical Genetics Part A, 1998, 79, 294-304.	2.4	116
21	Defects of blastogenesis. American Journal of Medical Genetics Part A, 2002, 115, 269-286.	2.4	104
22	Borderline normal intelligence in the Smith-Lemli-Opitz (RSH) syndrome. American Journal of Medical Genetics Part A, 1980, 5, 137-143.	2.4	103
23	RSH/SLO (â€œSmith-Lemli-Opitzâ€) syndrome: Historical, genetic, and developmental considerations. American Journal of Medical Genetics Part A, 1994, 50, 344-346.	2.4	89
24	Identification and characterization of a missense mutation in the O-linked Î²-N-acetylglucosamine (O-GlcNAc) transferase gene that segregates with X-linked intellectual disability. Journal of Biological Chemistry, 2017, 292, 8948-8963.	3.4	87
25	The Poland Syndrome â€“ Clinical and Genealogical Data, Dermatoglyphic Analysis, and Incidence. Human Heredity, 1973, 23, 97-104.	0.8	86
26	Amnion rupture sequence in previable fetuses. American Journal of Medical Genetics Part A, 1988, 31, 63-73.	2.4	86
27	G syndrome (hypertelorism with esophageal abnormality and hypospadias, or hypospadias-dysphagia,) Tj ETQq1 1 0.784314 rgBT /Ov Medical Genetics Part A, 1987, 28, 275-285.	2.4	82
28	Opitz G/BBB syndrome: Clinical comparisons of families linked to Xp22 and 22Q and a review of the literature. , 1996, 62, 305-317.		81
29	On the gates of hell and a most unusual gene. American Journal of Medical Genetics Part A, 1986, 23, 1-10.	2.4	77
30	Implications of malformations not due to amniotic bands in the amniotic band sequence. American Journal of Medical Genetics Part A, 1986, 24, 691-700.	2.4	76
31	Dubowitz syndrome: Review of 141 cases including 36 previously unreported patients. American Journal of Medical Genetics Part A, 1996, 63, 277-289.	2.4	76
32	Autosomal recessive syndrome of cerebellar ataxia and hypogonadotropic hypogonadism*. Clinical Genetics, 1975, 7, 426-434.	2.0	75
33	A new autosomal dominant acrofacial dysostosis syndrome. American Journal of Medical Genetics Part A, 1986, 25, 143-150.	2.4	72
34	Ovine ceroid-lipofuscinosis II: Pathologic changes interpreted in light of biochemical observations. American Journal of Medical Genetics Part A, 1988, 31, 159-170.	2.4	71
35	The perrault syndrome: Autosomal recessive ovarian dysgenesis with facultative, non-sex-limited sensorineural deafness. American Journal of Medical Genetics Part A, 1979, 4, 239-246.	2.4	68
36	Meier-Gorlin syndrome: Report of eight additional cases and review. American Journal of Medical Genetics Part A, 2001, 102, 115-124.	2.4	68

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37	Differential diagnosis of Nager acrofacial dysostosis syndrome: Report of four patients with Nager syndrome and discussion of other related syndromes. American Journal of Medical Genetics Part A, 1983, 14, 209-224.	2.4	66
38	The Perlman syndrome: Familial renal dysplasia with Wilms tumor, fetal gigantism and multiple congenital anomalies. American Journal of Medical Genetics Part A, 1984, 19, 195-207.	2.4	64
39	A novel germline PIGA mutation in Ferroâ€Cerebroâ€Cutaneous syndrome: A neurodegenerative Xâ€linked epileptic encephalopathy with systemic ironâ€overload. American Journal of Medical Genetics, Part A, 2014, 164, 17-28.	1.2	62
40	Polytopic anomalies with agenesis of the lower vertebral column. American Journal of Medical Genetics Part A, 1999, 87, 99-114.	2.4	61
41	I-Cell disease, mucolipidosis II. European Journal of Pediatrics, 1973, 114, 259-292.	2.7	60
42	Studies of malformation syndromes of man XXIX: The Wiedemann-Beckwith syndrome. European Journal of Nuclear Medicine and Molecular Imaging, 1976, 123, 139-166.	2.1	58
43	Editorial comment on the paper by Crowe and Dickerman: On congenital lymphedema. American Journal of Medical Genetics Part A, 1986, 24, 127-129.	2.4	58
44	FG syndrome update 1988: Note of 5 new patients and bibliography. American Journal of Medical Genetics Part A, 1988, 30, 309-328.	2.4	58
45	Further delineation of the C (trigonocephaly) syndrome. American Journal of Medical Genetics Part A, 1981, 9, 147-163.	2.4	56
46	Severe end of Opitz trigonocephaly (C) syndrome or new syndrome?. , 1999, 85, 438-446.		56
47	Dermatoglyphic traits as possible markers of developmental processes in humans. American Journal of Medical Genetics Part A, 1987, 26, 111-122.	2.4	50
48	Prenatal diagnosis of perinatally lethal osteogenesis imperfecta. American Journal of Medical Genetics Part A, 1983, 14, 353-359.	2.4	48
49	Mutations in CD96, a Member of the Immunoglobulin Superfamily, Cause a Form of the C (Opitz) Tj ETQq1 1 0.784314 rgBT /Overloc	6.2	48
50	A new X-linked mental retardation syndrome. American Journal of Medical Genetics Part A, 1985, 21, 697-705.	2.4	47
51	The Noonan syndrome. American Journal of Medical Genetics Part A, 1985, 21, 515-518.	2.4	43
52	Evidence for the ?midline? hypothesis in associated defects of laterality formation and multiple midline anomalies. American Journal of Medical Genetics Part A, 2001, 101, 382-387.	2.4	42
53	Tandem dup(1p) within the short arm of chromosome 1 in a child with ambiguous genitalia and multiple congenital anomalies. American Journal of Medical Genetics Part A, 1984, 17, 723-730.	2.4	40
54	CHOLESTEROL AND DEVELOPMENT: THE RSH ("SMITH-LEMLI-OPITZ") SYNDROME AND RELATED CONDITIONS. Fetal and Pediatric Pathology, 2002, 21, 153-181.	0.3	39

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55	Meckel on developmental pathology. American Journal of Medical Genetics, Part A, 2006, 140A, 115-128.	1.2	39
56	Syndrome of mental retardation, seizures, hypotonic cerebral palsy and megalocorneae, recessively inherited. European Journal of Pediatrics, 1975, 120, 1-18.	2.7	38
57	Editorial comment on the paper by de la Monte and Hutchins on familial polyasplenia. American Journal of Medical Genetics Part A, 1985, 21, 175-176.	2.4	37
58	Vaginal atresia (von Mayer-Rokitansky-KÃ¼ster or MRK anomaly) in hereditary renal adysplasia (HRA). American Journal of Medical Genetics Part A, 1987, 26, 873-876.	2.4	37
59	Nager â€œsyndromeâ€•versus â€œanomalyâ€•and its nosology with the postaxial acrofacial dysostosis syndrome of GenÃ©e and Wiedemann. American Journal of Medical Genetics Part A, 1987, 27, 959-963.	2.4	37
60	Klippel-Feil anomaly with Sprengel anomaly, omovertebral bone, thumb abnormalities, and flexion-crease changes: Novel association or syndrome?. American Journal of Medical Genetics Part A, 2001, 101, 158-162.	2.4	37
61	CHOLESTEROL AND DEVELOPMENT: THE RSH ("SMITH-LEMLI-OPITZ") SYNDROME AND RELATED CONDITIONS. Fetal and Pediatric Pathology, 2002, 21, 153-181.	0.3	37
62	Multiple meningiomas, craniofacial hyperostosis and retinal abnormalities in proteus syndrome. American Journal of Medical Genetics Part A, 2000, 93, 234-240.	2.4	36
63	Specific congenital heart defects in RSH/Smith-Lemli-Opitz syndrome: Postulated involvement of the Sonic Hedgehog pathway in syndromes with postaxial polydactyly or heterotaxia. Birth Defects Research Part A: Clinical and Molecular Teratology, 2003, 67, 149-153.	1.6	36
64	Autosomal recessive syndrome of pseudogliomatous blindness, osteoporosis and mild mental retardation. Clinical Genetics, 1976, 9, 324-332.	2.0	36
65	Autosomal dominant and sporadic radio-ulnar synostosis. , 1997, 68, 127-134.		35
66	Studies of malformation syndromes of humans XXXIIIC: The FG syndrome â€" further studies on three affected individuals from the FG family. American Journal of Medical Genetics Part A, 1982, 12, 147-154.	2.4	34
67	An evolutionary and developmental biology approach to gastroschisis. Birth Defects Research, 2019, 111, 294-311.	1.5	34
68	Studies of malformation syndromes of man XXXIX: A craniosynostosis-craniofacial dysostosis syndrome with mental retardation and other malformations: ?Craniofacial dyssynostosis?. European Journal of Nuclear Medicine and Molecular Imaging, 1976, 123, 15-28.	2.1	33
69	Brief historical note: The concept of â€œgonadal dysgenesisâ€• American Journal of Medical Genetics Part A, 1979, 4, 333-343.	2.4	32
70	Smith-Lemli-Opitz (RSH) syndrome bibliography. American Journal of Medical Genetics Part A, 1987, 28, 745-750.	2.4	32
71	An Xq22.3 duplication detected by comparative genomic hybridization microarray (Array-CGH) defines a new locus (FGS5) for FG syndrome. American Journal of Medical Genetics, Part A, 2005, 139A, 221-226.	1.2	32
72	Simpson-Golabi-Behmel syndrome: Follow-up of the michigan family. American Journal of Medical Genetics Part A, 1988, 30, 301-308.	2.4	31

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73	Associations and syndromes: Terminology in clinical genetics and birth defects epidemiology: Comments on Khoury, Moore, and Evans. American Journal of Medical Genetics Part A, 1994, 49, 14-20.	2.4	31
74	Kniest dysplasia: Radiologic, histopathological, and scanning electronmicroscopic findings. , 1996, 63, 34-45.		31
75	Studies of malformation syndromes of man XXXXIA: anatomical studies in the Hanhart syndrome ?A pathogenetic hypothesis. European Journal of Nuclear Medicine and Molecular Imaging, 1976, 122, 1-17.	2.1	30
76	Two sporadic cases of amelia/phocomelia with similar phenotype: Rare and unusually symmetrical form of FFU dysostosis or separate entity?. American Journal of Medical Genetics Part A, 1986, 25, 239-245.	2.4	30
77	Proximal femoral focal deficiency (PFFD) and fibular a/hypoplasia (FA/H): A model of a developmental field defect. American Journal of Medical Genetics Part A, 1995, 55, 427-432.	2.4	30
78	An X-linked recessive basal ganglia disorder with mental retardation. American Journal of Medical Genetics Part A, 1985, 21, 681-689.	2.4	29
79	Smith-Lemli-Opitz (RSH) syndrome bibliography: 1964-1993. American Journal of Medical Genetics Part A, 1994, 50, 339-343.	2.4	29
80	A De Novo Nonsense Mutation in MAGEL2 in a Patient Initially Diagnosed as Opitz-C: Similarities Between Schaaf-Yang and Opitz-C Syndromes. Scientific Reports, 2017, 7, 44138.	3.3	29
81	Comments on biological asymmetry. American Journal of Medical Genetics Part A, 2001, 101, 359-369.	2.4	28
82	Down syndrome: Comments and reflections on the 50th anniversary of Lejeune's discovery. American Journal of Medical Genetics, Part A, 2009, 149A, 2647-2654.	1.2	28
83	Cervical Ribs Are More Prevalent in Stillborn Fetuses than in Live-Born Infants and Are Strongly Associated with Fetal Aneuploidy. Pediatric and Developmental Pathology, 2011, 14, 431-437.	1.0	28
84	Studies of malformation syndromes of man XXXXIIB: Mother and son affected with the ulnar-mammary syndrome type Pallister. European Journal of Nuclear Medicine and Molecular Imaging, 1976, 123, 225-235.	2.1	24
85	Megacystis-microcolon-intestinal hypoperistalsis syndrome and aganglionosis in trisomy 18. American Journal of Medical Genetics Part A, 2001, 102, 293-296.	2.4	24
86	Hypertrichosis lanuginosa in a mother and son. Clinical Genetics, 1976, 10, 303-306.	2.0	24
87	A fetus with upper limb amelia, â€œcaudal regressionâ€•and Dandy-Walker defect with an insulin-depedent diabetic mother. European Journal of Pediatrics, 1980, 134, 139-143.	2.7	23
88	Unilateral partial tibia defect with preaxial polydactyly, general micromelia, and trigonomacrocephaly with a note on â€œdevelopmental resistanceâ€• American Journal of Medical Genetics Part A, 1983, 14, 467-471.	2.4	23
89	Perrault syndrome in sisters. American Journal of Medical Genetics Part A, 1985, 22, 629-631.	2.4	23
90	The GenÃ©e-Wiedemann syndrome, an acrofacial dysostosisâ€”further observation. American Journal of Medical Genetics Part A, 1987, 27, 971-975.	2.4	23

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91	Blaschkolinear malformation syndrome in complex trisomy-7 mosaicism. , 1999, 87, 375-383.		23
92	Heart development: An introduction. American Journal of Medical Genetics Part A, 2000, 97, 238-247.	2.4	23
93	FG syndrome: Linkage analysis in two families supporting a new gene localization at Xp22.3 [FGS3]. American Journal of Medical Genetics Part A, 2002, 112, 6-11.	2.4	23
94	Lethal Forms of Chondrodysplastic Dwarfism. Pediatrics, 1974, 53, 76-85.	2.1	23
95	Developmental termsâ€"some proposals: First report of an International working group. American Journal of Medical Genetics Part A, 1979, 3, 297-302.	2.4	22
96	Sedaghatian congenital lethal metaphyseal chondrodysplasiaâ€"observations in a second Iranian family and histopathological studies. American Journal of Medical Genetics Part A, 1987, 26, 583-590.	2.4	22
97	Reflections on the pathogenesis of Down syndrome. American Journal of Medical Genetics Part A, 2005, 37, 38-51.	2.4	22
98	Ulnar ray A/hypoplasia. Evidence for a developmental field defect on the basis of genetic heterogeneity. Report of three Brazilian families. American Journal of Medical Genetics Part A, 1986, 25, 195-206.	2.4	21
99	Severe case of al Awadi/Raas-Rothschild syndrome or new, possibly autosomal recessive facio-skeleto-genital syndrome. American Journal of Medical Genetics Part A, 1995, 56, 168-172.	2.4	21
100	Noonan's syndrome in the male. Journal of Pediatrics, 1965, 67, 936.	1.8	20
101	Hutterite cerebro-osteo-nephrodysplasia: Autosomal recessive trait in a Lehrerleut Hutterite family from Montana. American Journal of Medical Genetics Part A, 1985, 22, 521-529.	2.4	20
102	Developmental field theory and observationsâ€"accidental progress?. American Journal of Medical Genetics Part A, 1986, 25, 1-8.	2.4	20
103	Microcephaly, lymphedema, and chorioretinal dysplasia: Report of two additional cases. American Journal of Medical Genetics Part A, 1994, 53, 99-101.	2.4	19
104	Twenty-seven-year follow-up in the Wolf-Hirschhorn syndrome. American Journal of Medical Genetics Part A, 1995, 55, 459-461.	2.4	19
105	The evaluation of infants with the Zellweger (cerebroâ€hepatoâ€renal) syndrome*. Clinical Genetics, 1975, 7, 413-416.	2.0	19
106	Generalized gangliosidosis type II (Juvenile GM1 gangliosidosis). European Journal of Pediatrics, 1975, 120, 151-180.	2.7	18
107	Behavior phenotype of FG syndrome: Cognition, personality, and behavior in eleven affected boys. American Journal of Medical Genetics Part A, 2000, 97, 112-118.	2.4	18
108	MORTALITY AND PATHOLOGICAL FINDINGS IN C (OPITZ TRIGONOCEPHALY) SYNDROME. Fetal and Pediatric Pathology, 2006, 25, 211-231.	0.7	18

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109	Virilism as a late manifestation in the Bardet-Biedl syndrome. American Journal of Medical Genetics Part A, 1980, 7, 279-292.	2.4	17
110	Lincoln vs. Douglas again; comments on the papers by Curry et al, Greenberg et al, and Belmont et al. American Journal of Medical Genetics Part A, 1987, 26, 69-71.	2.4	17
111	Delayed Mutation as a Cause of Genetic Disease in Man: Achondroplasia and the Wiedemann-Beckwith Syndrome. , 1977, , 65-79.		17
112	An unusual dysplasia-malformation-cancer syndrome in two patients. American Journal of Medical Genetics Part A, 1978, 1, 279-289.	2.4	16
113	Heterogeneity and minor anomalies. , 2000, 91, 254-255.		16
114	Prenatal death in Smith-Lemli-Opitz/RSH syndrome. American Journal of Medical Genetics, Part A, 2005, 138A, 61-65.	1.2	16
115	The neurofaciodigitorenal (NFDR) syndrome. American Journal of Medical Genetics Part A, 1982, 11, 329-336.	2.4	15
116	Smith-Lemli-Opitz syndrome in Japan. American Journal of Medical Genetics Part A, 1998, 75, 118-119.	2.4	15
117	PRENATAL DEATH IN FRASER SYNDROME. Fetal and Pediatric Pathology, 2005, 24, 223-238.	0.7	15
118	The montana fetal genetic pathology program and a review of prenatal death in humans. American Journal of Medical Genetics Part A, 1987, 28, 93-112.	2.4	14
119	Disequilibrium syndrome in montana hutterites. American Journal of Medical Genetics Part A, 1985, 22, 567-569.	2.4	13
120	Craniocervical Anomalies in Dubowitz Syndrome. Pediatric Neurosurgery, 2003, 38, 238-243.	0.7	13
121	Screening of CD96 and ASXL1 in 11 patients with Opitz C or Bohringâ€“Opitz syndromes. American Journal of Medical Genetics, Part A, 2016, 170, 24-31.	1.2	13
122	Minamata disease. Acta Neuropathologica, 1973, 26, 275-284.	7.7	12
123	Heterogeneity of nonlethal severe short-limbed dwarfism. Journal of Pediatrics, 1977, 91, 918-923.	1.8	12
124	Clinicopathological conference: A 29-yr-old man with recurrent episodes of fever, abdominal pain, and vomiting. American Journal of Medical Genetics Part A, 1984, 18, 249-264.	2.4	12
125	The genetics of quantifiable homeostasis: I. The general issues. American Journal of Medical Genetics Part A, 1986, 24, 159-169.	2.4	12
126	Heterogeneity and minor anomalies. American Journal of Medical Genetics Part A, 2000, 92, 373-375.	2.4	12

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127	Sixty tears of X-linked mental retardation: A historical footnote. American Journal of Medical Genetics Part A, 2000, 97, 228-233.	2.4	12
128	The FG Syndromes (Online Mendelian Inheritance in Man 305450): Perspective in 2008. Advances in Pediatrics, 2008, 55, 123-170.	1.4	12
129	2011 William Allan Award: Development and Evolution 1. American Journal of Human Genetics, 2012, 90, 392-404.	6.2	12
130	Autopsy findings in a stillborn female infant with the Osebold-Remondini syndrome. American Journal of Medical Genetics Part A, 1985, 22, 811-819.	2.4	11
131	Bibliography on X-linked mental retardation and related subjects III (1986). American Journal of Medical Genetics Part A, 1986, 23, 69-99.	2.4	11
132	Developmental field theory and the molecular analysis of morphogenesis: A comment on Dr. Slavkin's observations. American Journal of Medical Genetics Part A, 1993, 47, 687-688.	2.4	11
133	Goethe's bone and the beginnings of morphology. American Journal of Medical Genetics Part A, 2004, 126A, 1-8.	2.4	11
134	Invited editorial comment: Further reflections on gastroschisis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2008, 148C, 192-198.	1.6	11
135	A deletion 13q34/duplication 14q32.2-14q32.33 syndrome diagnosed 50 years after neonatal presentation as infantile hypercalcemia. , 2011, 155, 833-839.		11
136	ADAM â€œsequenceâ€•part II: Hypothesis and speculation. American Journal of Medical Genetics, Part A, 2015, 167, 478-503.	1.2	11
137	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. Scientific Reports, 2018, 8, 694.	3.3	11
138	Another â€œnewâ€•form, the Palagonia type of acrofacial dysostosis in Sicilian family. American Journal of Medical Genetics Part A, 1997, 69, 388-394.	2.4	10
139	Opitz trigonocephaly syndrome presenting with sudden unexplained death in the operating room: a case report. Journal of Medical Case Reports, 2011, 5, 222.	0.8	10
140	Book reviewISCN 2013: An International System for Human Cytogenetic Nomenclature (2013). ShafferLG, McGowanâ€JordanJ, SchmidM, editors. Published in collaboration with Cytogenetic and Genome Research by Karger, Basel. VI and 140 pp. plus foldâ€•out, 11 Fig. 4 tables.. American Journal of Medical Genetics, Part A, 2013, 161, 2108-2109.	1.2	10
141	MENDEL: Morphologist and Mathematician Founder of Genetics â€“ To Begin a Celebration of the 2015 Sesquicentennial of Mendel's Presentation in 1865 of hisVersuche Ã¼ber Pflanzenhybriden. Molecular Genetics & Genomic Medicine, 2015, 3, 1-7.	1.2	10
142	Sudden death in childhood in a case of the G syndrome. American Journal of Medical Genetics Part A, 1987, 28, 293-296.	2.4	9
143	Development: Clinical and evolutionary considerations. American Journal of Medical Genetics, Part A, 2007, 143A, 2853-2861.	1.2	9
144	The N syndrome, a â€œnewâ€•multiple congenital anomaly-mental retardation syndrome:Studies of malformation syndromes in man XXVII. Clinical Genetics, 2008, 6, 237-246.	2.0	9

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145	Annals of morphology. Atavisms: Phylogenetic lazarus?. American Journal of Medical Genetics, Part A, 2013, 161, 2822-2835.	1.2	9
146	Johann Friedrich Meckel the Younger (1781-1833). American Journal of Medical Genetics Part A, 1984, 18, 571-586.	2.4	8
147	The first Meckel oration: On the causes of morphological differences in a population of guinea pigs. American Journal of Medical Genetics Part A, 1984, 18, 591-616.	2.4	8
148	Mandibulofacial dysostosis or bilateral hemifacial microsomia with hearing loss, telecanthus, tetramelic postaxial hexadactyly, congenital hypotonia and lymphedema with joint hypermobility, and pigmentary dysplasia: A new syndrome?. American Journal of Medical Genetics Part A, 1989, 33, 433-435.	2.4	8
149	Generalized lymphangiectasis associated with chylothorax; a possible dysplasia of the lymphatic system. European Journal of Pediatrics, 1974, 118, 9-24.	2.7	7
150	Historical perspective on developmental concepts and terminology. American Journal of Medical Genetics, Part A, 2013, 161, 2711-2725.	1.2	7
151	Annals of morphology THEODOR BOVERI (1862â€“1915) To commemorate the centenary of his death and contributions to the Suttonâ€“Boveri hypothesis. American Journal of Medical Genetics, Part A, 2016, 170, 2803-2829.	1.2	7
152	Clinicopathological conference: Renal failure with hypercalcemia, renal stones, multiple pathologic fractures, and growth failure. American Journal of Medical Genetics Part A, 1983, 14, 169-179.	2.4	6
153	Complete absence or deficiency of one half of the body. American Journal of Medical Genetics Part A, 1998, 76, 197-201.	2.4	6
154	Genitourinary Anomalies of Pediatric FG Syndrome. Journal of Urology, 2007, 178, 656-659.	0.4	6
155	Phenotypes, pleiotropy, and phylogeny. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 329-340.	1.6	6
156	Sudden infant death â€œsyndromeâ€â€”Insights and future directions from a Utah population database analysis. American Journal of Medical Genetics, Part A, 2017, 173, 177-182.	1.2	6
157	Encomium: Robert J. Gorlin. American Journal of Medical Genetics Part A, 1993, 46, 316-316.	2.4	5
158	Behold the CHILD. , 2000, 90, 336-338.		5
159	Heterogeneity of cardio-facio-cutaneous syndrome. American Journal of Medical Genetics Part A, 2000, 95, 144-144.	2.4	5
160	Human anotocephaly (aprosopus, acrania-synotia) in the Vilnius anatomical collection. American Journal of Medical Genetics Part A, 2001, 101, 163-171.	2.4	5
161	Invited comment: Gastroschisis. American Journal of Medical Genetics, Part A, 2007, 143A, 635-638.	1.2	5
162	Genetic caring. The professionalization of genetic services in the USA. American Journal of Medical Genetics Part A, 1979, 3, 1-5.	2.4	4

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163	Bibliography on X-linked mental retardation and related subjects II (1985). American Journal of Medical Genetics Part A, 1985, 21, 719-729.	2.4	4
164	Bibliography on X-linked mental retardation, the fragile X and related subjects IV (1988). American Journal of Medical Genetics Part A, 1988, 30, 31-60.	2.4	4
165	Studies of malformation syndromes of man XXXVIII: the BD syndrome. European Journal of Pediatrics, 1975, 120, 191-198.	2.7	3
166	David Klein: An appreciation. American Journal of Medical Genetics Part A, 1990, 37, 320-324.	2.4	3
167	Syndromal foramina parietalia permagna: ?new? or FG syndrome? Comments on the paper by Chrzanowska et al. [1998]. , 1998, 78, 406-407.		3
168	Skewed X chromosome inactivation in carriers is not a constant finding in FG syndrome. European Journal of Human Genetics, 2003, 11, 352-356.	2.8	3
169	Nuchal cystic hygroma in five fetuses from 1819 to 1826 in the Meckel-anatomical collections at the University of Halle, Germany. American Journal of Medical Genetics, Part A, 2007, 143A, 119-128.	1.2	3
170	The RSH/â€œSmithâ€“Lemliâ€“Opitzâ€•Syndrome: Historical footnote. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 242-249.	1.6	3
171	Serendipity or prepared mind? Recollections of the KOP translocation (1967) and of one form of Perrault syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 387-396.	1.6	3
172	Biographical noteâ€”Laurence H. Snyder. American Journal of Medical Genetics Part A, 1981, 8, 447-448.	2.4	2
173	Hans-Rudolf Wiedemann: An appreciation. American Journal of Medical Genetics Part A, 1992, 43, 737-739.	2.4	2
174	Meinhard Robinow: An appreciation. American Journal of Medical Genetics Part A, 1995, 59, 4-7.	2.4	2
175	Arthur G. Steinberg: An appreciation. American Journal of Medical Genetics Part A, 1995, 59, 245-249.	2.4	2
176	Previously apparently undescribed autosomal recessive MCA/MR syndrome with light fixation, retinal cone dystrophy, and seizures: The M syndrome. , 1999, 82, 194-198.		2
177	RE: Correspondence from Wieczorek & Gillessen-Kaesbach and Hing & Parisi. American Journal of Medical Genetics, Part A, 2006, 140A, 2385-2385.	1.2	2
178	Why is the construction: Hypoplastic left heart â€œsyndromeâ€•a misnomer? And: What is a syndrome, anyhow?. American Journal of Medical Genetics, Part A, 2011, 155, 360-362.	1.2	2
179	The power of stories in Pediatrics and Genetics. Italian Journal of Pediatrics, 2016, 42, 35.	2.6	2
180	Living history biography: An afterthought. American Journal of Medical Genetics, Part A, 2021, 185, 1355-1362.	1.2	2

#	Article	IF	Citations
181	Comments on biological asymmetry. American Journal of Medical Genetics Part A, 2001, 101, 359.	2.4	2
182	Rett syndrome bibliography. American Journal of Medical Genetics Part A, 1986, 25, 39-46.	2.4	1
183	Bibliography on X-linked mental retardation, the fragile X, and related subjects V (1991). American Journal of Medical Genetics Part A, 1991, 38, 173-185.	2.4	1
184	Familial broad terminal phalanges with one individual showing additional anomalies. , 1997, 71, 271-274.		1
185	Previously undescribed syndrome of spondylometaphyseal dysplasia, osteocartilaginous metaplasia of long bones, and progressive osteolysis of distal phalanges. , 1998, 80, 187-195.		1
186	Scott Rogers on dinosaur behavior, in: The Annals of Morphology. American Journal of Medical Genetics, Part A, 2005, 134A, 357-358.	1.2	1
187	A chicken consultation with ramifications. American Journal of Medical Genetics, Part A, 2005, 134A, 117-128.	1.2	1
188	Colophon: Vere dignum et justum estâ€‰â€¦â€‰An unedited MS. American Journal of Medical Genetics, Part A, 2006, 140A, 2150-2154.	1.2	1
189	An amnion implantation hypothesis: A conceptual framework for mechanismâ€based studies of amnion adhesion. American Journal of Medical Genetics, Part A, 2014, 164, 1092-1093.	1.2	1
190	Introductionâ€”A Pallister Jubilee. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 367-369.	1.6	1
191	To the Editor: Concerning Rajagopal MD etÂal.. Fetal and Pediatric Pathology, 2016, 35, 207-208.	0.7	1
192	Arno G. Motulsky, 1923â€“2018, Luck and Service. American Journal of Medical Genetics, Part A, 2018, 176, 1285-1288.	1.2	1
193	Megacystisâ€microcolonâ€intestinal hypoperistalsis syndrome and aganglionosis in trisomy 18. American Journal of Medical Genetics Part A, 2001, 102, 293-296.	2.4	1
194	Smith-Lemli-Opitz syndrome: clinical and biochemical findings in Brazilian patients. Scalco et al. Genetics and Molecular Biology (this issue). Genetics and Molecular Biology, 2006, 29, 437-438.	1.3	1
195	Editorial comment on the CPC by tripp et al: â€œFever and abdominal pain in a young woman with the down syndrome and eisenmenger complexâ€: Down syndrome and death of the mentally retarded. American Journal of Medical Genetics Part A, 1983, 14, 285-287.	2.4	0
196	Autosomal dominant recurrent encephalopathy of childhood. American Journal of Medical Genetics Part A, 1983, 15, 127-133.	2.4	0
197	A living past: Introduction to the section on history of the American Journal of Medical Genetics. American Journal of Medical Genetics Part A, 1991, 40, fmi-fmii.	2.4	0
198	In honor or JÃ¶rgen Spranger at 65. , 1996, 63, 1-2.		0

#	Article	IF	Citations
199	Encomium: Oswaldo Frota-Pessoa. American Journal of Medical Genetics Part A, 1996, 63, 581-584.	2.4	0
200	Timeo danaos . . .. , 1998, 78, 487-488.		0
201	James V. Neel. American Journal of Medical Genetics Part A, 1999, 83, 149-151.	2.4	0
202	Robert Layman Summitt, M.D. 1932-1998. , 1999, 86, 403-404.		0
203	The days of our age: TheAmerican Journal of Medical Genetics in the next millenium. , 2000, 90, 1-5.		0
204	Encomium and dedication: Angelo serra-four decades in human and medical genetics. American Journal of Medical Genetics Part A, 2005, 37, 1-8.	2.4	0
205	FRASER SYNDROME. Fetal and Pediatric Pathology, 2008, 27, 294-294.	0.7	0
206	The FG Syndrome from a Pathological Perspective. Fetal and Pediatric Pathology, 2011, 30, 71-76.	0.7	0
207	Response to Li and Liu's â€œDarwin's statements on reversion or atavismâ€• American Journal of Medical Genetics, Part A, 2014, 164, 2140-2140.	1.2	0
208	Remembered: F. Clarke Fraser. American Journal of Medical Genetics, Part A, 2015, 167, 2229-2230.	1.2	0
209	Of mice and cats (both calico): Mary F Lyon, FRS (1925â€“2014). American Journal of Medical Genetics, Part A, 2015, 167, 1957-1961.	1.2	0
210	Remembered: Elisabeth G. Kaveggia. American Journal of Medical Genetics, Part A, 2015, 167, 681-682.	1.2	0
211	An inner god: BEN E. KATZ (1921â€“2015) as geneticist. American Journal of Medical Genetics, Part A, 2015, 167, 2516-2519.	1.2	0
212	Comment: The midline. American Journal of Medical Genetics, Part A, 2015, 167, 2566-2567.	1.2	0
213	To the Editor: Concerning Rodriguez etÂal.. Fetal and Pediatric Pathology, 2015, 34, 340-340.	0.7	0
214	Editorial comment on McPherson and Cold. American Journal of Medical Genetics, Part A, 2016, 170, 60-61.	1.2	0
215	GAUDEAMUS IGITURâ€¦ In gratitude to John Carey for his stewardship of theAmerican Journal of Medical Genetics2001-2016. , 2016, 170, 2501-2502.		0
216	Annals of morphology fields and prepatterns. Editorial Festschrift for John C. Carey, MD, MPH. American Journal of Medical Genetics, Part A, 2016, 170, 2503-2522.	1.2	0

#	Article	IF	Citations
217	Second Pallisterâ€Opitz Genetics Symposium, Helena, Montana, July 2015. American Journal of Medical Genetics, Part A, 2016, 170, 1405-1421.	1.2	0
218	In gratitude for an undeserved gift*. American Journal of Medical Genetics, Part A, 2017, 173, 1147-1148.	1.2	0
219	Philip D. Pallister of Montana. American Journal of Medical Genetics, Part A, 2019, 179, 9-12.	1.2	0
220	The Work of Becerra-Solano et al. (2008) on Amniotic Disruption-Adhesion-Mutilation (ADAM or DAB) Sequence Deserves Comment. Fetal and Pediatric Pathology, 2019, 38, 91-92.	0.7	0
221	Jacqueline A. Noonan. American Journal of Medical Genetics, Part A, 2020, 182, 2820-2822.	1.2	0