

John M Opitz

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

214
papers

9,083
citations

47
h-index

90
g-index

238
ext. papers

10,060
ext. citations

4.9
avg, IF

5.51
L-index

#	Paper	IF	Citations
214	Living history biography: An afterthought. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1355-1362		1362
213	Jacqueline A. Noonan. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2820-2822	2.5	
212	Philip D. Pallister of Montana. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 9-12	2.5	
211	The Work of Becerra-Solano et al. (2008) on Amniotic Disruption-Adhesion-Mutilation (ADAM or DAB) Sequence Deserves Comment. <i>Fetal and Pediatric Pathology</i> , 2019 , 38, 91-92	1.7	
210	An evolutionary and developmental biology approach to gastroschisis. <i>Birth Defects Research</i> , 2019 , 111, 294-311	2.9	22
209	A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. <i>Scientific Reports</i> , 2018 , 8, 694	4.9	5
208	Arno G. Motulsky, 1923-2018, Luck and Service. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1285-1288	2.5	1
207	A De Novo Nonsense Mutation in MAGEL2 in a Patient Initially Diagnosed as Opitz-C: Similarities Between Schaaf-Yang and Opitz-C Syndromes. <i>Scientific Reports</i> , 2017 , 7, 44138	4.9	24
206	In gratitude for an undeserved gift. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1147-1148	2.5	
205	Identification and characterization of a missense mutation in the X-linked N-acetylglucosamine (-GlcNAc) transferase gene that segregates with X-linked intellectual disability. <i>Journal of Biological Chemistry</i> , 2017 , 292, 8948-8963	5.4	58
204	Phenotypes, pleiotropy, and phylogeny. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017 , 175, 329-340	3.1	5
203	Sudden infant death "syndrome"-Insights and future directions from a Utah population database analysis. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 177-182	2.5	5
202	GAUDEAMUS Igitur In gratitude to John Carey for his stewardship of the American Journal of Medical Genetics 2001-2016. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2501-2	2.5	
201	Annals of morphology fields and prepatterns. Editorial Festschrift for John C. Carey, MD, MPH. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2503-22	2.5	
200	Annals of morphology THEODOR BOVERI (1862-1915) To commemorate the centenary of his death and contributions to the Sutton-Boveri hypothesis 2016 , 170, 2803-2829		6
199	The power of stories in Pediatrics and Genetics. <i>Italian Journal of Pediatrics</i> , 2016 , 42, 35	3.2	0
198	Second Pallister-Opitz Genetics Symposium, Helena, Montana, July 2015. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 1405-21	2.5	

197	To the Editor: Concerning Rajagopal MD et'al. <i>Fetal and Pediatric Pathology</i> , 2016 , 35, 207-8	1.7	1
196	Screening of CD96 and ASXL1 in 11 patients with Opitz C or Bohring-Opitz syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 24-31	2.5	11
195	ADAM "sequence" part II: hypothesis and speculation. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 478-503	2.5	9
194	Disruptions of topological chromatin domains cause pathogenic rewiring of gene-enhancer interactions. <i>Cell</i> , 2015 , 161, 1012-1025	56.2	1207
193	To the Editor: Concerning Rodriguez et al. <i>Fetal and Pediatric Pathology</i> , 2015 , 34, 340	1.7	
192	Remembered: F. Clarke Fraser. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2229-30	2.5	
191	Of mice and cats (both calico): Mary F Lyon, FRS (1925-2014). <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 1957-61	2.5	
190	MENDEL: Morphologist and Mathematician Founder of Genetics - To Begin a Celebration of the 2015 Sesquicentennial of Mendel's Presentation in 1865 of his Versuche über Pflanzenhybriden. <i>Molecular Genetics & Genomic Medicine</i> , 2015 , 3, 1-7	2.3	6
189	Remembered: Elisabeth G. Kaveggia. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 681-2	2.5	
188	An inner god: BEN E. KATZ (1921-2015) as geneticist. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2516-9	2.5	
187	Comment: The midline. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2566-7	2.5	
186	Response to Li and Liu's "Darwin's statements on reversion or atavism". <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2140	2.5	
185	Introduction--a Pallister jubilee. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014 , 166C, 367-9	3.1	1
184	Serendipity or prepared mind? Recollections of the KOP translocation (1967) and of one form of Perrault syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014 , 166C, 387-96	3.1	3
183	A novel germline PIGA mutation in Ferro-Cerebro-Cutaneous syndrome: a neurodegenerative X-linked epileptic encephalopathy with systemic iron-overload. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 17-28	2.5	51
182	An amnion implantation hypothesis: a conceptual framework for mechanism-based studies of amnion adhesion. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1092-3	2.5	1
181	Book reviewISCN 2013: An International System for Human Cytogenetic Nomenclature (2013). Shaffer LG, McGowan-Jordan J, Schmid M, editors. Published in collaboration with Cytogenetic and Genome Research by Karger, Basel. VI and 140 pp. plus fold-out, 11 Fig. 4 tables. 2013 , 161, 2108-2109		8
180	Historical perspective on developmental concepts and terminology. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2711-25	2.5	6

179	Annals of morphology. Atavisms: phylogenetic Lazarus?. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2822-35	2.5	6
178	The RSH/"Smith-Lemli-Opitz" syndrome: historical footnote. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2012 , 160C, 242-9	3.1	3
177	Germline mutations in DIS3L2 cause the Perlman syndrome of overgrowth and Wilms tumor susceptibility. <i>Nature Genetics</i> , 2012 , 44, 277-84	36.3	173
176	2011 William Allan Award: development and evolution. <i>American Journal of Human Genetics</i> , 2012 , 90, 392-404	11	8
175	The FG syndrome from a pathological perspective. <i>Fetal and Pediatric Pathology</i> , 2011 , 30, 71-6	1.7	
174	Opitz trigonocephaly syndrome presenting with sudden unexplained death in the operating room: a case report. <i>Journal of Medical Case Reports</i> , 2011 , 5, 222	1.2	8
173	Why is the construction: Hypoplastic left heart "syndrome" a misnomer? And: What is a syndrome, anyhow?. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 360-2	2.5	2
172	A deletion 13q34/duplication 14q32.2-14q32.33 syndrome diagnosed 50 years after neonatal presentation as infantile hypercalcemia. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 833-9	2.5	9
171	Cervical ribs are more prevalent in stillborn fetuses than in live-born infants and are strongly associated with fetal aneuploidy. <i>Pediatric and Developmental Pathology</i> , 2011 , 14, 431-7	2.2	25
170	Mutations in mitochondrial histidyl tRNA synthetase HARS2 cause ovarian dysgenesis and sensorineural hearing loss of Perrault syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011 , 108, 6543-8	11.5	200
169	Down syndrome: comments and reflections on the 50th anniversary of Lejeune's discovery. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 2647-54	2.5	20
168	The FG syndromes (Online Mendelian Inheritance in Man 305450): perspective in 2008. <i>Advances in Pediatrics</i> , 2008 , 55, 123-70	2.2	12
167	Fraser syndrome. <i>Fetal and Pediatric Pathology</i> , 2008 , 27, 294	1.7	
166	Invited editorial comment: further reflections on gastroschisis. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2008 , 148C, 192-8	3.1	11
165	Genitourinary anomalies of pediatric FG syndrome. <i>Journal of Urology</i> , 2007 , 178, 656-9	2.5	5
164	Nuchal cystic hygroma in five fetuses from 1819 to 1826 in the Meckel-anatomical collections at the University of Halle, Germany. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 119-28	2.5	1
163	Invited comment: gastroschisis. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 635-8	2.5	5
162	Development: clinical and evolutionary considerations. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 2853-61	2.5	8

161	A recurrent mutation in MED12 leading to R961W causes Opitz-Kaveggia syndrome. <i>Nature Genetics</i> , 2007 , 39, 451-3	36.3	157
160	Mutations in CD96, a member of the immunoglobulin superfamily, cause a form of the C (Opitz trigonocephaly) syndrome. <i>American Journal of Human Genetics</i> , 2007 , 81, 835-41	11	44
159	Meckel on developmental pathology. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 115-28	2.5	25
158	Colophon: vere dignum et justum est ... an unedited MS. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 2150-4	2.5	1
157	RE: Correspondence from Wieczorek & Gillessen-Kaesbach and Hing & Parisi. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 2385	2.5	2
156	Mortality and pathological findings in C (Opitz trigonocephaly) syndrome. <i>Fetal and Pediatric Pathology</i> , 2006 , 25, 211-31	1.7	16
155	Smith-Lemli-Opitz syndrome: clinical and biochemical findings in Brazilian patients. Scalco et al. Genetics and Molecular Biology (this issue). <i>Genetics and Molecular Biology</i> , 2006 , 29, 437-438	2	1
154	A chicken consultation with ramifications. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 134A, 117-28		1
153	Prenatal death in Fraser syndrome. <i>Fetal and Pediatric Pathology</i> , 2005 , 24, 223-38	1.7	12
152	Encomium and dedication: Angelo serraour decades in human and medical genetics. <i>American Journal of Medical Genetics Part A</i> , 2005 , 37, 1-8		
151	Prenatal death in Smith-Lemli-Opitz/RSH syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 138, 61-5	2.5	15
150	Scott Rogers on dinosaur behavior, in: the annals of morphology. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 134, 357-8	2.5	0
149	An Xq22.3 duplication detected by comparative genomic hybridization microarray (Array-CGH) defines a new locus (FGS5) for FG syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 139, 221-6	2.5	26
148	Goethe's bone and the beginnings of morphology. <i>American Journal of Medical Genetics Part A</i> , 2004 , 126A, 1-8		6
147	Specific congenital heart defects in RSH/Smith-Lemli-Opitz syndrome: postulated involvement of the sonic hedgehog pathway in syndromes with postaxial polydactyly or heterotaxia. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2003 , 67, 149-53		32
146	Skewed X chromosome inactivation in carriers is not a constant finding in FG syndrome. <i>European Journal of Human Genetics</i> , 2003 , 11, 352-6	5.3	3
145	Craniocervical anomalies in Dubowitz syndrome. Three cases and a literature review. <i>Pediatric Neurosurgery</i> , 2003 , 38, 238-43	0.9	10
144	FG syndrome: linkage analysis in two families supporting a new gene localization at Xp22.3 [FGS3]. <i>American Journal of Medical Genetics Part A</i> , 2002 , 112, 6-11		22

143	Defects of blastogenesis. <i>American Journal of Medical Genetics Part A</i> , 2002 , 115, 269-86	95
142	Cholesterol and development: the RSH ("Smith-Lemli-Opitz") syndrome and related conditions. <i>Fetal and Pediatric Pathology</i> , 2002 , 21, 153-81	26
141	CHOLESTEROL AND DEVELOPMENT: THE RSH ("SMITH-LEMLI-OPITZ") SYNDROME AND RELATED CONDITIONS. <i>Fetal and Pediatric Pathology</i> , 2002 , 21, 153-181	34
140	Klippel-Feil anomaly with Sprengel anomaly, omovertebral bone, thumb abnormalities, and flexion-crease changes: Novel association or syndrome?. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 158-162	32
139	Comments on biological asymmetry. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 359-369	24
138	Evidence for the midline hypothesis in associated defects of laterality formation and multiple midline anomalies. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 382-387	36
137	Human anotocephaly (aprosopus, acrania-synotia) in the Vilnius anatomical collection. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 163-71	4
136	Meier-Gorlin syndrome: report of eight additional cases and review. <i>American Journal of Medical Genetics Part A</i> , 2001 , 102, 115-24	58
135	Megacystis-microcolon-intestinal hypoperistalsis syndrome and aganglionosis in trisomy 18. <i>American Journal of Medical Genetics Part A</i> , 2001 , 102, 293-296	16
134	Comments on biological asymmetry 2001 , 101, 359	2
133	Megacystis-microcolon-intestinal hypoperistalsis syndrome and aganglionosis in trisomy 18 2001 , 102, 293	1
132	Behold the CHILD. <i>American Journal of Medical Genetics Part A</i> , 2000 , 90, 336-8	4
131	Heterogeneity and minor anomalies. <i>American Journal of Medical Genetics Part A</i> , 2000 , 91, 254-5	8
130	Multiple meningiomas, craniofacial hyperostosis and retinal abnormalities in Proteus syndrome. <i>American Journal of Medical Genetics Part A</i> , 2000 , 93, 234-40	30
129	Heterogeneity of cardio-facio-cutaneous syndrome. <i>American Journal of Medical Genetics Part A</i> , 2000 , 95, 144	4
128	Behavior phenotype of FG syndrome: cognition, personality, and behavior in eleven affected boys. <i>American Journal of Medical Genetics Part A</i> , 2000 , 97, 112-8	16
127	Sixty years of X-linked mental retardation: a historical footnote. <i>American Journal of Medical Genetics Part A</i> , 2000 , 97, 228-33	9
126	Heart development: An introduction. <i>American Journal of Medical Genetics Part A</i> , 2000 , 97, 238-247	19

125	Previously apparently undescribed autosomal recessive MCA/MR syndrome with light fixation, retinal cone dystrophy, and seizures: the M syndrome. <i>American Journal of Medical Genetics Part A</i> , 1999 , 82, 194-8	1
124	James V. Neel. <i>American Journal of Medical Genetics Part A</i> , 1999 , 83, 149-151	
123	Severe end of Opitz trigonocephaly (C) syndrome or new syndrome?. <i>American Journal of Medical Genetics Part A</i> , 1999 , 85, 438-46	48
122	Robert Layman Summitt, M.D. 1932-1998. <i>American Journal of Medical Genetics Part A</i> , 1999 , 86, 403-4	
121	Polytopic anomalies with agenesis of the lower vertebral column. <i>American Journal of Medical Genetics Part A</i> , 1999 , 87, 99-114	54
120	Blaschkolinear malformation syndrome in complex trisomy-7 mosaicism. <i>American Journal of Medical Genetics Part A</i> , 1999 , 87, 375-83	20
119	Smith-Lemli-Opitz syndrome in Japan. <i>American Journal of Medical Genetics Part A</i> , 1998 , 75, 118-119	14
118	Complete absence or deficiency of one half of the body. <i>American Journal of Medical Genetics Part A</i> , 1998 , 76, 197-201	6
117	Errors of morphogenesis and developmental field theory 1998 , 76, 291-296	105
116	Syndromal foramina parietalia permagna: BewŁbr FG syndrome? Comments on the paper by Chrzanowska et al. [1998] 1998 , 78, 406-407	3
115	Timeo danaos. <i>American Journal of Medical Genetics Part A</i> , 1998 , 78, 487-8	
114	The syndromes of Sotos and Weaver: reports and review. <i>American Journal of Medical Genetics Part A</i> , 1998 , 79, 294-304	90
113	Previously undescribed syndrome of spondylometaphyseal dysplasia, osteocartilaginous metaplasia of long bones, and progressive osteolysis of distal phalanges. <i>American Journal of Medical Genetics Part A</i> , 1998 , 80, 187-95	0
112	Opitz G/BBB syndrome, a defect of midline development, is due to mutations in a new RING finger gene on Xp22. <i>Nature Genetics</i> , 1997 , 17, 285-91	36.3 296
111	Autosomal dominant and sporadic radio-ulnar synostosis 1997 , 68, 127-134	27
110	Another "new" form, the palagonia type of acrofacial dysostosis in a Sicilian family. <i>American Journal of Medical Genetics Part A</i> , 1997 , 69, 388-94	8
109	Familial broad terminal phalanges with one individual showing additional anomalies 1997 , 71, 271-274	1
108	Evaluation of mental retardation: recommendations of a Consensus Conference: American College of Medical Genetics. <i>American Journal of Medical Genetics Part A</i> , 1997 , 72, 468-77	272

107	Resynthesizing evolutionary and developmental biology. <i>Developmental Biology</i> , 1996 , 173, 357-72	3.1	502
106	Opitz G/BBB syndrome: clinical comparisons of families linked to Xp22 and 22q, and a review of the literature. <i>American Journal of Medical Genetics Part A</i> , 1996 , 62, 305-17		66
105	Festschrift in honor of Jürgen Spranger. <i>American Journal of Medical Genetics Part A</i> , 1996 , 63, 1-327		
104	Dubowitz syndrome: review of 141 cases including 36 previously unreported patients. <i>American Journal of Medical Genetics Part A</i> , 1996 , 63, 277-89		64
103	Kniest dysplasia: radiologic, histopathological, and scanning electronmicroscopic findings. <i>American Journal of Medical Genetics Part A</i> , 1996 , 63, 34-45		21
102	Encomium: Oswaldo Frota-Pessoa. <i>American Journal of Medical Genetics Part A</i> , 1996 , 63, 581-4		
101	Proximal femoral focal deficiency (PFFD) and fibular A/hypoplasia (FA/H): a model of a developmental field defect. <i>American Journal of Medical Genetics Part A</i> , 1995 , 55, 427-32		24
100	Twenty-seven-year follow-up in the Wolf-Hirschhorn syndrome. <i>American Journal of Medical Genetics Part A</i> , 1995 , 55, 459-61		14
99	Severe case of Al Awadi/Raas-Rothschild syndrome or new, possibly autosomal recessive facio-skeleto-genital syndrome. <i>American Journal of Medical Genetics Part A</i> , 1995 , 56, 168-72		20
98	Meinhard Robinow: an appreciation. <i>American Journal of Medical Genetics Part A</i> , 1995 , 59, 4-7		1
97	Arthur G. Steinberg: an appreciation. <i>American Journal of Medical Genetics Part A</i> , 1995 , 59, 245-9		2
96	Smith-Lemli-Opitz (RSH) syndrome bibliography: 1964-1993. <i>American Journal of Medical Genetics Part A</i> , 1994 , 50, 339-43		26
95	RSH/SLO ("Smith-Lemli-Opitz") syndrome: historical, genetic, and developmental considerations. <i>American Journal of Medical Genetics Part A</i> , 1994 , 50, 344-6		79
94	Microcephaly, lymphedema, and chorioretinal dysplasia: report of two additional cases. <i>American Journal of Medical Genetics Part A</i> , 1994 , 53, 99-101		18
93	Encomium: Robert J. Gorlin. <i>American Journal of Medical Genetics Part A</i> , 1993 , 46, 316-316		4
92	Developmental field theory and the molecular analysis of morphogenesis: A comment on Dr. Slavkin's observations. <i>American Journal of Medical Genetics Part A</i> , 1993 , 47, 687-688		10
91	Hans-Rudolf Wiedemann: an appreciation. <i>American Journal of Medical Genetics Part A</i> , 1992 , 43, 737-9		1
90	Bibliography on X-linked mental retardation, the fragile X, and related Xp22 subjects V (1991). <i>American Journal of Medical Genetics Part A</i> , 1991 , 38, 173-85		1

89	Reflections on the pathogenesis of Down syndrome. <i>American Journal of Medical Genetics Part A</i> , 1990 , 7, 38-51	9
88	David Klein: an appreciation. <i>American Journal of Medical Genetics Part A</i> , 1990 , 37, 320-4	3
87	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?. <i>American Journal of Medical Genetics Part A</i> , 1989 , 33, 433-5	6
86	Bibliography on X-linked mental retardation, the fragile X and related subjects IV (1988). <i>American Journal of Medical Genetics Part A</i> , 1988 , 30, 31-60	3
85	Simpson-Golabi-Behmel syndrome: follow-up of the Michigan family. <i>American Journal of Medical Genetics Part A</i> , 1988 , 30, 301-8	28
84	FG syndrome update 1988: note of 5 new patients and bibliography. <i>American Journal of Medical Genetics Part A</i> , 1988 , 30, 309-28	53
83	Amnion rupture sequence in previable fetuses. <i>American Journal of Medical Genetics Part A</i> , 1988 , 31, 63-73	74
82	Ovine ceroid-lipofuscinosis II: Pathologic changes interpreted in light of biochemical observations. <i>American Journal of Medical Genetics Part A</i> , 1988 , 5, 159-70	70
81	Smith-Lemli-Opitz syndrome-type II: multiple congenital anomalies with male pseudohermaphroditism and frequent early lethality. <i>American Journal of Medical Genetics Part A</i> , 1987 , 26, 45-57	200
80	Lincoln vs. Douglas again; comments on the papers by Curry et al, Greenberg et al, and Belmont et al. <i>American Journal of Medical Genetics Part A</i> , 1987 , 26, 69-71	17
79	Dermatoglyphic traits as possible markers of developmental processes in humans. <i>American Journal of Medical Genetics Part A</i> , 1987 , 26, 111-22	46
78	Sedaghatian congenital lethal metaphyseal chondrodysplasia--observations in a second Iranian family and histopathological studies. <i>American Journal of Medical Genetics Part A</i> , 1987 , 26, 583-90	18
77	Vaginal atresia (von Mayer-Rokitansky-Kuster or MRK anomaly) in hereditary renal adysplasia (HRA). <i>American Journal of Medical Genetics Part A</i> , 1987 , 26, 873-6	34
76	Isochromosome 12p mosaicism (Pallister mosaic aneuploidy or Pallister-Killian syndrome): report of 11 cases. <i>American Journal of Medical Genetics Part A</i> , 1987 , 27, 257-74	123
75	Nager "syndrome" versus "anomaly" and its nosology with the postaxial acrofacial dysostosis syndrome of GenB and Wiedemann. <i>American Journal of Medical Genetics Part A</i> , 1987 , 27, 959-63	24
74	The GenB-Wiedemann syndrome, an acrofacial dysostosis--further observation. <i>American Journal of Medical Genetics Part A</i> , 1987 , 27, 971-5	21
73	G syndrome (hypertelorism with esophageal abnormality and hypospadias, or hypospadias-dysphagia, or "Opitz-Frias" or "Opitz-G" syndrome)--perspective in 1987 and bibliography. <i>American Journal of Medical Genetics Part A</i> , 1987 , 28, 275-85	73
72	Sudden death in childhood in a case of the G syndrome. <i>American Journal of Medical Genetics Part A</i> , 1987 , 28, 293-6	8

71	Smith-Lemli-Opitz (RSH) syndrome bibliography. <i>American Journal of Medical Genetics Part A</i> , 1987 , 28, 745-50	28
70	The Montana Fetal Genetic Pathology Program and a review of prenatal death in humans. <i>American Journal of Medical Genetics Part A</i> , 1987 , 3, 93-112	10
69	Bibliography of X-linked mental retardation and related subjects. III. (1986). <i>American Journal of Medical Genetics Part A</i> , 1986 , 23, 69-99	10
68	The genetics of quantifiable homeostasis: I. The general issues. <i>American Journal of Medical Genetics Part A</i> , 1986 , 24, 159-69	11
67	Implications of malformations not due to amniotic bands in the amniotic band sequence. <i>American Journal of Medical Genetics Part A</i> , 1986 , 24, 691-700	69
66	Rett syndrome bibliography. <i>American Journal of Medical Genetics Part A</i> , 1986 , 1, 39-46	
65	Rett syndrome at an institution for the developmentally disabled. <i>American Journal of Medical Genetics Part A</i> , 1986 , 1, 85-97	120
64	Developmental field theory and observations—accidental progress?. <i>American Journal of Medical Genetics Part A</i> , 1986 , 25, 1-8	17
63	The DiGeorge anomaly as a developmental field defect. <i>American Journal of Medical Genetics Part A</i> , 1986 , 2, 113-27	154
62	A new autosomal dominant acrofacial dysostosis syndrome. <i>American Journal of Medical Genetics Part A</i> , 1986 , 2, 143-50	68
61	Ulnar ray a/hypoplasia: evidence for a developmental field defect on the basis of genetic heterogeneity. Report of three Brazilian families. <i>American Journal of Medical Genetics Part A</i> , 1986 , 2, 195-206	21
60	Fibular a/hypoplasia: review and documentation of the fibular developmental field. <i>American Journal of Medical Genetics Part A</i> , 1986 , 2, 215-38	117
59	Two sporadic cases of amelia/phocomelia with similar phenotype: rare and unusually symmetrical form of FFU dysostosis or separate entity?. <i>American Journal of Medical Genetics Part A</i> , 1986 , 2, 239-45	29
58	The developmental field concept. <i>American Journal of Medical Genetics Part A</i> , 1985 , 21, 1-11	108
57	Editorial comment on the paper by de la Monte and Hutchins on familial polyasplenia. <i>American Journal of Medical Genetics Part A</i> , 1985 , 21, 175-176	36
56	Noonan syndrome: a review. <i>American Journal of Medical Genetics Part A</i> , 1985 , 21, 493-506	367
55	The Noonan syndrome. <i>American Journal of Medical Genetics Part A</i> , 1985 , 21, 515-8	32
54	An X-linked recessive basal ganglia disorder with mental retardation. <i>American Journal of Medical Genetics Part A</i> , 1985 , 21, 681-9	26

53	A new X-linked mental retardation syndrome. <i>American Journal of Medical Genetics Part A</i> , 1985 , 21, 697-705	43
52	Bibliography on X-linked mental retardation and related subjects II (1985). <i>American Journal of Medical Genetics Part A</i> , 1985 , 21, 719-29	4
51	The Brachmann-de Lange syndrome. <i>American Journal of Medical Genetics Part A</i> , 1985 , 22, 89-102	141
50	Hutterite cerebro-osteo-nephrodysplasia: autosomal recessive trait in a Lehrerleut Hutterite family from Montana. <i>American Journal of Medical Genetics Part A</i> , 1985 , 22, 521-9	17
49	Disequilibrium syndrome in Montana Hutterites. <i>American Journal of Medical Genetics Part A</i> , 1985 , 22, 567-9	12
48	Perrault syndrome in sisters. <i>American Journal of Medical Genetics Part A</i> , 1985 , 22, 629-31	23
47	Autopsy findings in a stillborn female infant with the Osebold-Remondini syndrome. <i>American Journal of Medical Genetics Part A</i> , 1985 , 22, 811-9	10
46	Tandem dup (1p) within the short arm of chromosome 1 in a child with ambiguous genitalia and multiple congenital anomalies. <i>American Journal of Medical Genetics Part A</i> , 1984 , 17, 723-30	38
45	Clinicopathological conference: a 29-yr-old man with recurrent episodes of fever, abdominal pain, and vomiting. <i>American Journal of Medical Genetics Part A</i> , 1984 , 18, 249-64	11
44	Johann Friedrich Meckel the Younger (1781-1833). <i>American Journal of Medical Genetics Part A</i> , 1984 , 18, 571-86	6
43	The first Meckel oration: on the causes of morphological differences in a population of guinea pigs. <i>American Journal of Medical Genetics Part A</i> , 1984 , 18, 591-616	7
42	The Perlman syndrome: familial renal dysplasia with Wilms tumor, fetal gigantism and multiple congenital anomalies. <i>American Journal of Medical Genetics Part A</i> , 1984 , 19, 195-207	56
41	Renal failure with hypercalcemia, renal stones, multiple pathologic fractures, and growth failure. <i>American Journal of Medical Genetics Part A</i> , 1983 , 14, 169-79	4
40	Differential diagnosis of Nager acrofacial dysostosis syndrome: report of four patients with Nager syndrome and discussion of other related syndromes. <i>American Journal of Medical Genetics Part A</i> , 1983 , 14, 209-24	61
39	Editorial comment on the CPC by Tripp et al: "Fever and abdominal pain in a young woman with the Down syndrome and Eisenmenger complex": Down syndrome and death of the mentally retarded. <i>American Journal of Medical Genetics Part A</i> , 1983 , 14, 285-7	
38	Prenatal diagnosis of perinatally lethal osteogenesis imperfecta. <i>American Journal of Medical Genetics Part A</i> , 1983 , 14, 353-9	39
37	Brief clinical report: unilateral partial tibia defect with preaxial polydactyly, general micromelia, and trigonmacrocephaly with a note on "developmental resistance". <i>American Journal of Medical Genetics Part A</i> , 1983 , 14, 467-71	22
36	Autosomal dominant recurrent encephalopathy of childhood. <i>American Journal of Medical Genetics Part A</i> , 1983 , 15, 127-33	

- 35 The neurofaciodigitorenal (NFDR) syndrome. *American Journal of Medical Genetics Part A*, **1982**, 11, 329-36 13
- 34 Studies of malformation syndromes of humans XXXIIC: the FG syndrome - further studies on three affected individuals from the FG family. *American Journal of Medical Genetics Part A*, **1982**, 12, 147-54 34
- 33 CNS anomalies and the midline as a "developmental field". *American Journal of Medical Genetics Part A*, **1982**, 12, 443-55 240
- 32 Biographical note--Laurence H. Snyder. *American Journal of Medical Genetics Part A*, **1981**, 8, 447-8 2
- 31 Further delineation of the C (trigonocephaly) syndrome. *American Journal of Medical Genetics Part A*, **1981**, 9, 147-63 50
- 30 A fetus with upper limb amelia, "caudal regression" and Dandy-Walker defect with an insulin-dependent diabetic mother. *European Journal of Pediatrics*, **1980**, 134, 139-43 4.1 22
- 29 Borderline normal intelligence in the Smith-Lemli-Opitz (RSH) syndrome. *American Journal of Medical Genetics Part A*, **1980**, 5, 137-43 91
- 28 Virilism as a late manifestation in the Bardet-Biedl syndrome. *American Journal of Medical Genetics Part A*, **1980**, 7, 279-92 14
- 27 Genetic caring. the professionalization of genetic services in the USA. *American Journal of Medical Genetics Part A*, **1979**, 3, 1-5 4
- 26 Developmental terms -- some proposals: first report of an international working group. *American Journal of Medical Genetics Part A*, **1979**, 3, 297-302 20
- 25 The Perrault syndrome: autosomal recessive ovarian dysgenesis with facultative, non-sex-limited sensorineural deafness. *American Journal of Medical Genetics Part A*, **1979**, 4, 239-46 64
- 24 Brief historical note: the concept of "gonadal dysgenesis". *American Journal of Medical Genetics Part A*, **1979**, 4, 333-43 30
- 23 An unusual dysplasia-malformation-cancer syndrome in two patients. *American Journal of Medical Genetics Part A*, **1978**, 1, 279-89 15
- 22 Heterogeneity of nonlethal severe short-limbed dwarfism. *Journal of Pediatrics*, **1977**, 91, 918-23 3.6 11
- 21 Delayed Mutation as a Cause of Genetic Disease in Man: Achondroplasia and the Wiedemann-Beckwith Syndrome **1977**, 65-79 14
- 20 Hypertrichosis lanuginosa in a mother and son. *Clinical Genetics*, **1976**, 10, 303-6 4 22
- 19 Autosomal recessive syndrome of pseudogliomantous blindness, osteoporosis and mild mental retardation. *Clinical Genetics*, **1976**, 9, 324-32 4 32
- 18 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, 225-35 21

17	Studies of malformation syndromes of man XXXXIA: anatomical studies in the Hanhart syndrome--a pathogenetic hypothesis. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 1976 , 122, 1-17		27
16	Studies of malformation syndromes of man XXIX: the Wiedemann-Beckwith syndrome. Clinical, genetic and pathogenetic studies of 12 cases. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 1976 , 123, 139-66		57
15	Studies of malformation syndromes of man XXXIX: a craniostosis-craniofacial dysostosis syndrome with mental retardation and other malformations: "craniofacial dyssynostosis". <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 1976 , 123, 15-28		31
14	The evaluation of infants with the Zellweger (cerebro-hepato-renal) syndrome. <i>Clinical Genetics</i> , 1975 , 7, 413-6	4	14
13	Autosomal recessive syndrome of cerebellar ataxia and hypogonadotropic hypogonadism. <i>Clinical Genetics</i> , 1975 , 7, 426-34	4	59
12	Generalized gangliosidosis type II (juvenile GM1 gangliosidosis). A pathological, histochemical and ultrastructural study. <i>European Journal of Pediatrics</i> , 1975 , 120, 151-80	4.1	15
11	Studies of malformation syndromes of man XXXVIII: The BD syndrome. A "new" multiple congenital anomalies/mental retardation syndrome with athetoid cerebral palsy. <i>European Journal of Pediatrics</i> , 1975 , 120, 191-8	4.1	3
10	Syndrome of mental retardation, seizures, hypotonic cerebral palsy and megalocorneae, recessively inherited. <i>European Journal of Pediatrics</i> , 1975 , 120, 1-18	4.1	33
9	Studies of malformation syndromes in man. XXVII. The N syndrome, a "new" multiple congenital anomaly-mental retardation syndrome. <i>Clinical Genetics</i> , 1974 , 6, 237-46	4	8
8	Studies of malformation syndromes of man 33: the FG syndrome. An X-linked recessive syndrome of multiple congenital anomalies and mental retardation. <i>European Journal of Pediatrics</i> , 1974 , 117, 1-18 ^{4.1}	4.1	122
7	Generalized lymphangiectosis associated with chylothorax; a possible dysplasia of the lymphatic system. <i>European Journal of Pediatrics</i> , 1974 , 118, 9-24	4.1	6
6	Lethal Forms of Chondrodysplastic Dwarfism. <i>Pediatrics</i> , 1974 , 53, 76-85	7.4	7
5	I-Cell disease, mucopolipidosis II. <i>European Journal of Pediatrics</i> , 1973 , 114, 259-292	4.1	51
4	Minamata disease. A case report and a comparative study. <i>Acta Neuropathologica</i> , 1973 , 26, 275-84	14.3	12
3	The Poland syndrome-clinical and genealogical data, dermatoglyphic analysis, and incidence. <i>Human Heredity</i> , 1973 , 23, 97-104	1.1	76
2	Noonan's syndrome in the male. <i>Journal of Pediatrics</i> , 1965 , 67, 936	3.6	16
1	A NEWLY RECOGNIZED SYNDROME OF MULTIPLE CONGENITAL ANOMALIES. <i>Journal of Pediatrics</i> , 1964 , 64, 210-7	3.6	579