

A Micheil Innes

List of Publications by Year in descending order

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Version: 2024-02-01

135
papers

8,153
citations

57681

46
h-index

64407

83
g-index

143
all docs

143
docs citations

143
times ranked

16179
citing authors

#	ARTICLE	IF	CITATIONS
1	Outcome of over 1500 matches through the Matchmaker Exchange for rare disease gene discovery: The 2-year experience of Care4Rare Canada. <i>Genetics in Medicine</i> , 2022, 24, 100-108.	1.1	15
2	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. <i>American Journal of Human Genetics</i> , 2022, 109, 518-532.	2.6	8
3	Phenotypic spectrum of the recurrent <i>TRPM3</i> p.(Val837Met) substitution in seven individuals with global developmental delay and hypotonia. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1667-1675.	0.7	8
4	<i>Hnrnpul1</i> controls transcription, splicing, and modulates skeletal and limb development in vivo. <i>G3: Genes, Genomes, Genetics</i> , 2022, 12, .	0.8	3
5	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in <i>POLR3A</i> , <i>POLR3B</i> , and <i>POLR1C</i> . <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e660-e674.	1.8	26
6	<i>CHRNB1</i> -associated congenital myasthenia syndrome: Expanding the clinical spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 827-835.	0.7	6
7	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	0.7	17
8	A dyadic approach to the delineation of diagnostic entities in clinical genomics. <i>American Journal of Human Genetics</i> , 2021, 108, 8-15.	2.6	71
9	The de novo <i>CACNA1A</i> pathogenic variant Y1384C associated with hemiplegic migraine, early onset cerebellar atrophy and developmental delay leads to a loss of Cav2.1 channel function. <i>Molecular Brain</i> , 2021, 14, 27.	1.3	12
10	De novo <i>TRIM8</i> variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2021, 108, 357-367.	2.6	14
11	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1649-1665.	0.7	34
12	<i>TAOK1</i> is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	1.1	26
13	<i>ANKRD11</i> variants: <i>KBG</i> syndrome and beyond. <i>Clinical Genetics</i> , 2021, 100, 187-200.	1.0	21
14	Fifty years of recognizable patterns of human malformation: Insights and opportunities. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2653-2669.	0.7	7
15	Loss of function mutations in <i>GEMIN5</i> cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	5.8	28
16	Heterozygous <i>ANKRD17</i> loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	2.6	17
17	Nucleocytoplasmic transport of the RNA-binding protein <i>CELF2</i> regulates neural stem cell fates. <i>Cell Reports</i> , 2021, 35, 109226.	2.9	16
18	Clustered mutations in the <i>GRIK2</i> kainate receptor subunit gene underlie diverse neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2021, 108, 1692-1709.	2.6	18

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19	Response to Hamosh et al. American Journal of Human Genetics, 2021, 108, 1809-1810.	2.6	0
20	De novo variants in MPP5 cause global developmental delay and behavioral changes. Human Molecular Genetics, 2020, 29, 3388-3401.	1.4	5
21	Congenital hiatal hernia segregating with a duplication in 9q22.31q22.32 in two families. American Journal of Medical Genetics, Part A, 2020, 182, 3040-3047.	0.7	1
22	Skeletal Phenotypes Due to Abnormalities in Mitochondrial Protein Homeostasis and Import. International Journal of Molecular Sciences, 2020, 21, 8327.	1.8	8
23	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
24	A diagnostic approach to syndromic retinal dystrophies with intellectual disability. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 538-570.	0.7	7
25	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
26	Automated syndrome diagnosis by three-dimensional facial imaging. Genetics in Medicine, 2020, 22, 1682-1693.	1.1	47
27	When to think outside the autozygome: Best practices for exome sequencing in consanguineous families. Clinical Genetics, 2020, 97, 835-843.	1.0	11
28	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	3.8	121
29	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. American Journal of Human Genetics, 2020, 106, 143-152.	2.6	30
30	Overlapping phenotypes between SHORT and Noonan syndromes in patients with PTPN11 pathogenic variants. Clinical Genetics, 2020, 98, 10-18.	1.0	9
31	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	1.4	45
32	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	1.7	20
33	Neuropathy due to impaired axonal transport of non-fragmented mitochondria in MYH14 mutation carriers – Authors' reply. EBioMedicine, 2019, 49, 25.	2.7	2
34	Adult MTM1-related myopathy carriers. Neurology, 2019, 93, e1535-e1542.	1.5	18
35	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	1.4	32
36	The R941L mutation in MYH14 disrupts mitochondrial fission and associates with peripheral neuropathy. EBioMedicine, 2019, 45, 379-392.	2.7	37

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37	The value of diagnostic testing for parents of children with rare genetic diseases. <i>Genetics in Medicine</i> , 2019, 21, 2798-2806.	1.1	31
38	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. <i>Cell</i> , 2019, 177, 32-37.	13.5	113
39	39th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2018 Annual Meeting. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 674-746.	0.7	4
40	Biallelic sequence variants in INTS1 in patients with developmental delays, cataracts, and craniofacial anomalies. <i>European Journal of Human Genetics</i> , 2019, 27, 582-593.	1.4	23
41	<i>PNPT1</i> is a mitochondrial disease gene causing skeletal dysplasia, cataracts, and white matter changes. <i>Life Science Alliance</i> , 2019, 2, e201900353.	1.3	41
42	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1128-1136.	0.7	55
43	Unsolved recognizable patterns of human malformation: Challenges and opportunities. , 2018, 178, 382-386.		13
44	Clinical and genetic heterogeneity in Dubowitz syndrome: Implications for diagnosis, management and further research. , 2018, 178, 387-397.		15
45	BAFopathies™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. <i>Nature Communications</i> , 2018, 9, 4885.	5.8	83
46	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	5.8	70
47	Is <i>PNPT1</i> -related hearing loss ever non-syndromic? Whole exome sequencing of adult siblings expands the natural history of <i>PNPT1</i> -related disorders. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2487-2493.	0.7	13
48	<i>HCN1</i> mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. <i>Brain</i> , 2018, 141, 3160-3178.	3.7	96
49	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. <i>American Journal of Human Genetics</i> , 2018, 103, 786-793.	2.6	17
50	Primary brain calcification: an international study reporting novel variants and associated phenotypes. <i>European Journal of Human Genetics</i> , 2018, 26, 1462-1477.	1.4	48
51	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogyriposis. <i>European Journal of Human Genetics</i> , 2018, 26, 1752-1758.	1.4	32
52	Cover Image, Volume 176A, Number 5, May 2018. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, .	0.7	0
53	Phenotypic spectrum of Au-Kline syndrome: a report of six new cases and review of the literature. <i>European Journal of Human Genetics</i> , 2018, 26, 1272-1281.	1.4	26
54	Next-Generation Sequencing Using a Cardiac Gene Panel in Prenatally Diagnosed Cardiac Anomalies. <i>Journal of Obstetrics and Gynaecology Canada</i> , 2018, 40, 1417-1423.	0.3	2

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55	Mosaic trisomy 1q: a recurring chromosome anomaly that is a diagnostic challenge and is associated with a Fryns-like phenotype. <i>Prenatal Diagnosis</i> , 2017, 37, 602-610.	1.1	5
56	Response to correspondence of NDUFS4-related Leigh syndrome in Hutterites. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1452-1452.	0.7	0
57	When One Diagnosis Is Not Enough. <i>New England Journal of Medicine</i> , 2017, 376, 83-85.	13.9	27
58	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. <i>American Journal of Human Genetics</i> , 2017, 101, 139-148.	2.6	45
59	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	9.4	251
60	A novel NDUFS4 frameshift mutation causes Leigh disease in the Hutterite population. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 596-600.	0.7	14
61	Expansion of the <sc>GLE1</sc>-associated arthrogryposis multiplex congenita clinical spectrum. <i>Clinical Genetics</i> , 2017, 91, 426-430.	1.0	15
62	SHORT syndrome due to a novel de novo mutation in PRKCE (Protein Kinase C ϵ) impairing TORC2-dependent AKT activation. <i>Human Molecular Genetics</i> , 2017, 26, 3713-3721.	1.4	22
63	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 912-928.	3.9	160
64	A relatively mild skeletal ciliopathy phenotype consistent with cranioectodermal dysplasia is associated with a homozygous nonsynonymous mutation in <i>WDR35</i>. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 760-765.	0.7	10
65	Copy-number variations are enriched for neurodevelopmental genes in children with developmental coordination disorder. <i>Journal of Medical Genetics</i> , 2016, 53, 812-819.	1.5	40
66	Recommendations for the integration of genomics into clinical practice. <i>Genetics in Medicine</i> , 2016, 18, 1075-1084.	1.1	125
67	Development of a diagnostic <sc>DNA</sc> chip to screen for 30 autosomal recessive disorders in the Hutterite population. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 312-321.	0.6	5
68	The Role of <i>PIEZO2</i> in Human Mechanosensation. <i>New England Journal of Medicine</i> , 2016, 375, 1355-1364.	13.9	293
69	Expansion of phenotype and genotypic data in CRB2-related syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1436-1444.	1.4	36
70	A novel mutation in two Hmong families broadens the range of <i>STRA6</i>-related malformations to include contractures and camptodactyly. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 11-18.	0.7	15
71	GeneMatcher Aids in the Identification of a New Malformation Syndrome with Intellectual Disability, Unique Facial Dysmorphisms, and Skeletal and Connective Tissue Abnormalities Caused by De Novo Variants in <i>HNRNPK</i>. <i>Human Mutation</i> , 2015, 36, 1009-1014.	1.1	56
72	Matching Two Independent Cohorts Validates <i>DPH1</i> as a Gene Responsible for Autosomal Recessive Intellectual Disability with Short Stature, Craniofacial, and Ectodermal Anomalies. <i>Human Mutation</i> , 2015, 36, 1015-1019.	1.1	32

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73	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. American Journal of Human Genetics, 2015, 97, 894-903.	2.6	242
74	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	2.6	171
75	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	2.6	101
76	CODAS Syndrome Is Associated with Mutations of LONP1, Encoding Mitochondrial AAA+ Lon Protease. American Journal of Human Genetics, 2015, 96, 121-135.	2.6	127
77	Biallelic Mutations in <i>BRCA1</i> Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
78	Homozygous mutations in <i>MFN2</i> cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
79	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
80	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy. <i>Genes</i> . Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
81	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	1.4	101
82	Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities. European Journal of Medical Genetics, 2015, 58, 503-508.	0.7	15
83	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. American Journal of Human Genetics, 2015, 97, 862-868.	2.6	36
84	Next Generation Diagnostics for Rare Neurological Diseases: The Future is Here. Canadian Journal of Neurological Sciences, 2014, 41, 299-300.	0.3	1
85	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebrocostomandibular syndrome. Nature Communications, 2014, 5, 4483.	5.8	57
86	Refinement of the critical region of 1q41q42 microdeletion syndrome identifies <i>FBXO28</i> as a candidate causative gene for intellectual disability and seizures. American Journal of Medical Genetics, Part A, 2014, 164, 441-448.	0.7	24
87	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101
88	Neuropathologic Features of Pontocerebellar Hypoplasia Type 6. Journal of Neuropathology and Experimental Neurology, 2014, 73, 1009-1025.	0.9	28
89	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	2.6	77
90	High frequency of copy number variations (CNVs) in the chromosome 11p15 region in patients with Beckwith-Wiedemann syndrome. Human Genetics, 2014, 133, 321-330.	1.8	50

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91	A Peroxisomal Disorder of Severe Intellectual Disability, Epilepsy, and Cataracts Due to Fatty Acyl-CoA Reductase 1 Deficiency. <i>American Journal of Human Genetics</i> , 2014, 95, 602-610.	2.6	106
92	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 95, 227-234.	2.6	92
93	De novo exon 1 missense mutations of <i>SKI</i> and Shprintzen-Goldberg syndrome: Two new cases and a clinical review. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 676-684.	0.7	16
94	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	1.4	120
95	Titinopathy in a Canadian Family Sharing the British Founder Haplotype. <i>Canadian Journal of Neurological Sciences</i> , 2014, 41, 90-94.	0.3	5
96	Intellectual disability associated with a homozygous missense mutation in THOC6. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 62.	1.2	48
97	Homozygous Lamin A/C familial lipodystrophy R482Q mutation in autosomal recessive Emery Dreifuss muscular dystrophy. <i>Neuromuscular Disorders</i> , 2013, 23, 265-268.	0.3	23
98	Identification of Novel Mutations Confirms <i>Pde4d</i> as a Major Gene Causing Acrodysostosis. <i>Human Mutation</i> , 2013, 34, 97-102.	1.1	49
99	Mutations in PIK3R1 Cause SHORT Syndrome. <i>American Journal of Human Genetics</i> , 2013, 93, 158-166.	2.6	156
100	Recessive TRAPPC11 Mutations Cause a Disease Spectrum of Limb Girdle Muscular Dystrophy and Myopathy with Movement Disorder and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2013, 93, 181-190.	2.6	98
101	The hutterite variant of treacher collins syndrome: A 28-year-old story solved. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2855-2859.	0.7	7
102	A shared founder mutation underlies restrictive dermopathy in Old Colony (Dutch-German) Mennonite and Hutterite patients in North America. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1229-1232.	0.7	14
103	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. <i>Nature Genetics</i> , 2012, 44, 934-940.	9.4	621
104	Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 925-933.	2.6	188
105	GPSM2 Mutations Cause the Brain Malformations and Hearing Loss in Chudley-McCullough Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 1088-1093.	2.6	103
106	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. <i>American Journal of Human Genetics</i> , 2011, 89, 713-730.	2.6	178
107	Clinical, pathological, and molecular analyses of cardiovascular abnormalities in Costello syndrome: A Ras/MAPK pathway syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 486-507.	0.7	99
108	BBS genotype-phenotype assessment of a multiethnic patient cohort calls for a revision of the disease definition. <i>Human Mutation</i> , 2011, 32, 610-619.	1.1	100

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109	Intracerebral hemorrhage in a young man. <i>Cmaj</i> , 2011, 183, E61-E64.	0.9	8
110	A novel autosomal recessive malformation syndrome associated with developmental delay and distinctive facies maps to 16p tel in the Hutterite population. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1349-1356.	0.7	12
111	Expanding the phenotypic spectrum of lupus erythematosus in Aicardi-Goutières syndrome. <i>Arthritis and Rheumatism</i> , 2010, 62, 1469-1477.	6.7	183
112	Interstitial deletion of 11q in a mother and fetus: implications of directly transmitted chromosomal imbalances for prenatal genetic counseling. <i>Prenatal Diagnosis</i> , 2009, 29, 283-286.	1.1	10
113	Prenatal features of Costello syndrome: ultrasonographic findings and atrial tachycardia. <i>Prenatal Diagnosis</i> , 2009, 29, 682-690.	1.1	52
114	Mutations in the Heparan-Sulfate Proteoglycan Glypican 6 (GPC6) Impair Endochondral Ossification and Cause Recessive Omodysplasia. <i>American Journal of Human Genetics</i> , 2009, 84, 760-770.	2.6	106
115	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. <i>Journal of Medical Genetics</i> , 2009, 46, 598-606.	1.5	194
116	Unique disease heritage of the Dutch-German Mennonite population. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1072-1087.	0.7	38
117	Costello syndrome associated with novel germline <i>HRAS</i> mutations: An attenuated phenotype?. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 683-690.	0.7	61
118	An undiagnosed cytogenetic abnormality results in the misidentification of a Duchenne muscular dystrophy carrier. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1067-1071.	0.7	0
119	Clinical genetics and the Hutterite population: A review of Mendelian disorders. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1088-1098.	0.7	44
120	Origin and outcome of pregnancies affected by androgenetic/biparental chimerism. <i>Human Reproduction</i> , 2007, 22, 1114-1122.	0.4	75
121	Molecular genetic testing and genetic counseling. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2007, 87, 517-531.	1.0	0
122	Meckel syndrome in the Hutterite population is actually a Joubert-related cerebello-oculo-renal syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1715-1725.	0.7	20
123	Frequency of genomic rearrangements involving the SHFM3 locus at chromosome 10q24 in syndromic and non-syndromic split-hand/foot malformation. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1375-1383.	0.7	22
124	Re: 3C (Ritscher-Schinzel) syndrome: the importance of ruling out a terminal 6p deletion. <i>Clinical Dysmorphology</i> , 2005, 14, 209-210.	0.1	3
125	A locus for Bowen-Conradi syndrome maps to chromosome region 12p13.3. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 136-143.	0.7	12
126	Re: 3C (Ritscher-Schinzel) syndrome: the importance of ruling out a terminal 6p deletion. <i>Clinical Dysmorphology</i> , 2005, 14, 209-210.	0.1	0

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127	G Protein-Coupled Receptor-Dependent Development of Human Frontal Cortex. <i>Science</i> , 2004, 303, 2033-2036.	6.0	498
128	Bilateral frontoparietal polymicrogyria: Clinical and radiological features in 10 families with linkage to chromosome 16. <i>Annals of Neurology</i> , 2003, 53, 596-606.	2.8	120
129	A rational approach to the child with mental retardation for the paediatrician. <i>Paediatrics and Child Health</i> , 2003, 8, 345-356.	0.3	13
130	Bowen-Conradi syndrome in non Hutterite infant. <i>Clinical Dysmorphology</i> , 2002, 11, 147-148.	0.1	4
131	Third case of cerebral, ocular, dental, auricular, skeletal anomalies (CODAS) syndrome, further delineating a new malformation syndrome: First report of an affected male and review of literature. <i>American Journal of Medical Genetics Part A</i> , 2001, 102, 44-47.	2.4	17
132	Rhabdomyosarcoma in a Patient With Cardiofaciocutaneous Syndrome. <i>The American Journal of Pediatric Hematology/oncology</i> , 2000, 22, 546.	1.3	7
133	Genetic landmarks through philately - Henry Louis "Lou" Gehrig and amyotrophic lateral sclerosis. <i>Clinical Genetics</i> , 1999, 56, 425-427.	1.0	2
134	X-chromosome inactivation is mostly random in placental tissues of female monozygotic twins and triplets. , 1996, 61, 209-215.		30
135	Some perinatal characteristics of monozygotic twins who are dichorionic. <i>American Journal of Medical Genetics Part A</i> , 1995, 55, 71-76.	2.4	58