

# A Micheil Innes

## List of Publications by Year in descending order

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135  
papers

8,153  
citations

50170

46  
h-index

56606

83  
g-index

143  
all docs

143  
docs citations

143  
times ranked

15008  
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	G Protein-Coupled Receptor-Dependent Development of Human Frontal Cortex. <i>Science</i> , 2004, 303, 2033-2036.	6.0	498
3	The Role of <i>PIEZO2</i> in Human Mechanosensation. <i>New England Journal of Medicine</i> , 2016, 375, 1355-1364.	13.9	293
4	Biallelic Mutations in <i>BRCA1</i> Cause a New Fanconi Anemia Subtype. <i>Cancer Discovery</i> , 2015, 5, 135-142.	7.7	251
5	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	9.4	251
6	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. <i>American Journal of Human Genetics</i> , 2015, 97, 894-903.	2.6	242
7	Mutations in <i>DDX3X</i> Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
8	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy. <i>Nature Cell Biology</i> , 2015, 17, 1074-1087.	4.6	215
9	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of <i>EHMT1</i> haploinsufficiency to the core phenotype. <i>Journal of Medical Genetics</i> , 2009, 46, 598-606.	1.5	194
10	Haploinsufficiency of <i>SF3B4</i> , 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
11	Expanding the phenotypic spectrum of lupus erythematosus in Aicardi-Goutières syndrome. <i>Arthritis and Rheumatism</i> , 2010, 62, 1469-1477.	6.7	183
12	<i>TMEM237</i> Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the <i>TMEM</i> Family at the Ciliary Transition Zone. <i>American Journal of Human Genetics</i> , 2011, 89, 713-730.	2.6	178
13	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene <i>SLC39A8</i> . <i>American Journal of Human Genetics</i> , 2015, 97, 886-893.	2.6	171
14	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
15	Mutations in <i>PIK3R1</i> Cause SHORT Syndrome. <i>American Journal of Human Genetics</i> , 2013, 93, 158-166.	2.6	156
16	CODAS Syndrome Is Associated with Mutations of <i>LONP1</i> , Encoding Mitochondrial AAA+ Lon Protease. <i>American Journal of Human Genetics</i> , 2015, 96, 121-135.	2.6	127
17	Recommendations for the integration of genomics into clinical practice. <i>Genetics in Medicine</i> , 2016, 18, 1075-1084.	1.1	125
18	Pathogenic <i>DDX3X</i> Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. <i>Neuron</i> , 2020, 106, 404-420.e8.	3.8	121

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19	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
20	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
21	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. <i>Cell</i> , 2019, 177, 32-37.	13.5	113
22	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
23	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
24	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
25	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. <i>Genome Biology</i> , 2014, 15, R53.	13.9	101
26	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	2.6	101
27	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. <i>European Journal of Human Genetics</i> , 2015, 23, 1473-1481.	1.4	101
28	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
29	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
30	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
31	<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
32	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
33	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
34	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. <i>American Journal of Human Genetics</i> , 2014, 94, 73-79.	2.6	77
35	Origin and outcome of pregnancies affected by androgenetic/biparental chimerism. <i>Human Reproduction</i> , 2007, 22, 1114-1122.	0.4	75
36	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

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37	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
38	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
39	Homozygous mutations in <i>MFN2</i> cause multiple symmetric lipomatosis associated with neuropathy. <i>Human Molecular Genetics</i> , 2015, 24, 5109-5114.	1.4	61
40	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
41	Disrupted auto-regulation of the spliceosomal gene <i>SNRPB</i> causes cerebro-osteo-mandibular syndrome. <i>Nature Communications</i> , 2014, 5, 4483.	5.8	57
42	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
43	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1128-1136.	0.7	55
44	Prenatal features of Costello syndrome: ultrasonographic findings and atrial tachycardia. <i>Prenatal Diagnosis</i> , 2009, 29, 682-690.	1.1	52
45	High frequency of copy number variations (CNVs) in the chromosome 11p15 region in patients with Beckwith-Wiedemann syndrome. <i>Human Genetics</i> , 2014, 133, 321-330.	1.8	50
46	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
47	Intellectual disability associated with a homozygous missense mutation in <i>THOC6</i> . <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 62.	1.2	48
48	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
49	Automated syndrome diagnosis by three-dimensional facial imaging. <i>Genetics in Medicine</i> , 2020, 22, 1682-1693.	1.1	47
50	<i>WDR26</i> 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
51	De novo substitutions of <i>TRPM3</i> cause intellectual disability and epilepsy. <i>European Journal of Human Genetics</i> , 2019, 27, 1611-1618.	1.4	45
52	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
53	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020, 6, .	4.7	43
54	<i>PISD</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

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55	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
56	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
57	The R941L mutation in MYH14 disrupts mitochondrial fission and associates with peripheral neuropathy. <i>EBioMedicine</i> , 2019, 45, 379-392.	2.7	37
58	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. <i>American Journal of Human Genetics</i> , 2015, 97, 862-868.	2.6	36
59	Expansion of phenotype and genotypic data in CRB2-related syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1436-1444.	1.4	36
60	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
61	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
62	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogryposis. <i>European Journal of Human Genetics</i> , 2018, 26, 1752-1758.	1.4	32
63	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. <i>European Journal of Human Genetics</i> , 2019, 27, 738-746.	1.4	32
64	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
65	X-chromosome inactivation is mostly random in placental tissues of female monozygotic twins and triplets. <i>Am J Hum Genet</i> , 1996, 61, 209-215.		30
66	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. <i>American Journal of Human Genetics</i> , 2020, 106, 143-152.	2.6	30
67	Neuropathologic Features of Pontocerebellar Hypoplasia Type 6. <i>Journal of Neuropathology and Experimental Neurology</i> , 2014, 73, 1009-1025.	0.9	28
68	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	5.8	28
69	When One Diagnosis Is Not Enough. <i>New England Journal of Medicine</i> , 2017, 376, 83-85.	13.9	27
70	Phenotypic spectrum of Aukline 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
71	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
72	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	1.1	26

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73	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1338-1347.	1.1	25
74	Refinement of the critical region of 1q41q42 microdeletion syndrome identifies <i>FBXO28</i> as a candidate causative gene for intellectual disability and seizures. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 441-448.	0.7	24
75	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
76	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
77	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
78	SHORT syndrome due to a novel de novo mutation in PRKCE (Protein Kinase C $\epsilon$ ) impairing TORC2-dependent AKT activation. <i>Human Molecular Genetics</i> , 2017, 26, 3713-3721.	1.4	22
79	<i>ANKRD11</i> variants: KBG syndrome and beyond. <i>Clinical Genetics</i> , 2021, 100, 187-200.	1.0	21
80	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
81	Biallelic <i>CACNA2D2</i> variants in epileptic encephalopathy and cerebellar atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1395-1406.	1.7	20
82	Adult MTM1-related myopathy carriers. <i>Neurology</i> , 2019, 93, e1535-e1542.	1.5	18
83	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2021, 108, 1692-1709.	2.6	18
84	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
85	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
86	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
87	Heterozygous ANKRD17 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
88	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
89	Nucleocytoplasmic transport of the RNA-binding protein CELF2 regulates neural stem cell fates. <i>Cell Reports</i> , 2021, 35, 109226.	2.9	16
90	Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities. <i>European Journal of Medical Genetics</i> , 2015, 58, 503-508.	0.7	15

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91	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
92	Expansion of the <i>GLE1</i> -associated arthrogryposis multiplex congenita clinical spectrum. <i>Clinical Genetics</i> , 2017, 91, 426-430.	1.0	15
93	Clinical and genetic heterogeneity in Dubowitz syndrome: Implications for diagnosis, management and further research. , 2018, 178, 387-397.		15
94	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
95	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
96	A novel <i>NDUFS4</i> 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
97	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
98	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
99	Unsolved recognizable patterns of human malformation: Challenges and opportunities. , 2018, 178, 382-386.		13
100	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
101	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
102	A novel autosomal recessive malformation syndrome associated with developmental delay and distinctive facies maps to 16ptel in the Hutterite population. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1349-1356.	0.7	12
103	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
104	When to think outside the autozygome: Best practices for exome sequencing in "consanguineous" families. <i>Clinical Genetics</i> , 2020, 97, 835-843.	1.0	11
105	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
106	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
107	Overlapping phenotypes between SHORT and Noonan syndromes in patients with <i>PTPN11</i> pathogenic variants. <i>Clinical Genetics</i> , 2020, 98, 10-18.	1.0	9
108	Intracerebral hemorrhage in a young man. <i>Cmaj</i> , 2011, 183, E61-E64.	0.9	8

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109	Skeletal Phenotypes Due to Abnormalities in Mitochondrial Protein Homeostasis and Import. International Journal of Molecular Sciences, 2020, 21, 8327.	1.8	8
110	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. American Journal of Human Genetics, 2022, 109, 518-532.	2.6	8
111	Phenotypic spectrum of the recurrent <i>TRPM3</i> p.(Val837Met) substitution in seven individuals with global developmental delay and hypotonia. American Journal of Medical Genetics, Part A, 2022, 188, 1667-1675.	0.7	8
112	The Hutterite variant of Treacher Collins syndrome: A 28-year-old story solved. American Journal of Medical Genetics, Part A, 2013, 161, 2855-2859.	0.7	7
113	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
114	Fifty years of recognizable patterns of human malformation: Insights and opportunities. American Journal of Medical Genetics, Part A, 2021, 185, 2653-2669.	0.7	7
115	Rhabdomyosarcoma in a Patient With Cardiofaciocutaneous Syndrome. The American Journal of Pediatric Hematology/Oncology, 2000, 22, 546.	1.3	7
116	CHRNB1-associated congenital myasthenia syndrome: Expanding the clinical spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 827-835.	0.7	6
117	Titinopathy in a Canadian Family Sharing the British Founder Haplotype. Canadian Journal of Neurological Sciences, 2014, 41, 90-94.	0.3	5
118	Development of a diagnostic DNA chip to screen for 30 autosomal recessive disorders in the Hutterite population. Molecular Genetics & Genomic Medicine, 2016, 4, 312-321.	0.6	5
119	Mosaic trisomy 1q: a recurring chromosome anomaly that is a diagnostic challenge and is associated with a Frys-like phenotype. Prenatal Diagnosis, 2017, 37, 602-610.	1.1	5
120	De novo variants in <i>MPP5</i> cause global developmental delay and behavioral changes. Human Molecular Genetics, 2020, 29, 3388-3401.	1.4	5
121	Bowen-Conradi syndrome in non-Hutterite infant. Clinical Dysmorphology, 2002, 11, 147-148.	0.1	4
122	39th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2018 Annual Meeting. American Journal of Medical Genetics, Part A, 2019, 179, 674-746.	0.7	4
123	Re: 3C (Ritscher-Schinzel) syndrome: the importance of ruling out a terminal 6p deletion. Clinical Dysmorphology, 2005, 14, 209-210.	0.1	3
124	Hnrnpul1 controls transcription, splicing, and modulates skeletal and limb development in vivo. G3: Genes, Genomes, Genetics, 2022, 12, .	0.8	3
125	Genetic landmarks through philately - Henry Louis "Lou" Gehrig and amyotrophic lateral sclerosis. Clinical Genetics, 1999, 56, 425-427.	1.0	2
126	Neuropathy due to impaired axonal transport of non-fragmented mitochondria in MYH14 mutation carriers - Authors reply. EBioMedicine, 2019, 49, 25.	2.7	2



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127	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
128	Next Generation Diagnostics for Rare Neurological Diseases: The Future is Here. <i>Canadian Journal of Neurological Sciences</i> , 2014, 41, 299-300.	0.3	1
129	Congenital hiatal hernia segregating with a duplication in 9q22.31q22.32 in two families. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 3040-3047.	0.7	1
130	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
131	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
132	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
133	Cover Image, Volume 176A, Number 5, May 2018. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, .	0.7	0
134	Response to Hamosh et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1809-1810.	2.6	0
135	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