A Micheil Innes

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2337719/publications.pdf

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	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	9.4	621
2	G Protein-Coupled Receptor-Dependent Development of Human Frontal Cortex. Science, 2004, 303, 2033-2036.	6.0	498
3	The Role of PIEZO2 in Human Mechanosensation. New England Journal of Medicine, 2016, 375, 1355-1364.	13.9	293
4	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
5	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	9.4	251
6	SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. American Journal of Human Genetics, 2015, 97, 894-903.	2.6	242
7	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
8	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
9	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. Journal of Medical Genetics, 2009, 46, 598-606.	1.5	194
10	Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. American Journal of Human Genetics, 2012, 90, 925-933.	2.6	188
11	Expanding the phenotypic spectrum of lupus erythematosus in Aicardiâ€GoutiÃ¨res syndrome. Arthritis and Rheumatism, 2010, 62, 1469-1477.	6.7	183
12	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 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 SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	2.6	171
14	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. Journal of Clinical Investigation, 2017, 127, 912-928.	3.9	160
15	Mutations in PIK3R1 Cause SHORT Syndrome. American Journal of Human Genetics, 2013, 93, 158-166.	2.6	156
16	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
17	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	1.1	125
18	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	3.8	121

#	Article	IF	Citations
19	Bilateral frontoparietal polymicrogyria: Clinical and radiological features in 10 families with linkage to chromosome 16. Annals of Neurology, 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. Human Molecular Genetics, 2014, 23, 2888-2900.	1.4	120
21	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
22	Mutations in the Heparan-Sulfate Proteoglycan Glypican 6 (GPC6) Impair Endochondral Ossification and Cause Recessive Omodysplasia. American Journal of Human Genetics, 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. American Journal of Human Genetics, 2014, 95, 602-610.	2.6	106
24	GPSM2 Mutations Cause the Brain Malformations and Hearing Loss in Chudley-McCullough Syndrome. American Journal of Human Genetics, 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. Genome Biology, 2014, 15, R53.	13.9	101
26	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	2.6	101
27	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
28	BBS genotype-phenotype assessment of a multiethnic patient cohort calls for a revision of the disease definition. Human Mutation, 2011, 32, 610-619.	1.1	100
29	Clinical, pathological, and molecular analyses of cardiovascular abnormalities in Costello syndrome: A Ras/MAPK pathway syndrome. American Journal of Medical Genetics, Part A, 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. American Journal of Human Genetics, 2013, 93, 181-190.	2.6	98
31	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	3.7	96
32	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. American Journal of Human Genetics, 2014, 95, 227-234.	2.6	92
33	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	5.8	83
34	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
35	Origin and outcome of pregnancies affected by androgenetic/biparental chimerism. Human Reproduction, 2007, 22, 1114-1122.	0.4	75
36	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	2.6	71

#	Article	IF	Citations
37	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	5.8	70
38	Costello syndrome associated with novel germline HRAS mutations: An attenuated phenotype?. American Journal of Medical Genetics, Part A, 2008, 146A, 683-690.	0.7	61
39	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
40	Some perinatal characteristics of monozygotic twins who are dichorionic. American Journal of Medical Genetics Part A, 1995, 55, 71-76.	2.4	58
41	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebroâ€“costoâ€“mandibular syndrome. Nature Communications, 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 inHNRNPK. Human Mutation, 2015, 36, 1009-1014.	1.1	56
43	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	0.7	55
44	Prenatal features of Costello syndrome: ultrasonographic findings and atrial tachycardia. Prenatal Diagnosis, 2009, 29, 682-690.	1.1	52
45	High frequency of copy number variations (CNVs) in the chromosome 11p15 region in patients with Beckwithâ€“Wiedemann syndrome. Human Genetics, 2014, 133, 321-330.	1.8	50
46	Identification of Novel Mutations ConfirmsPde4das a Major Gene Causing Acrodysostosis. Human Mutation, 2013, 34, 97-102.	1.1	49
47	Intellectual disability associated with a homozygous missense mutation in THOC6. Orphanet Journal of Rare Diseases, 2013, 8, 62.	1.2	48
48	Primary brain calcification: an international study reporting novel variants and associated phenotypes. European Journal of Human Genetics, 2018, 26, 1462-1477.	1.4	48
49	Automated syndrome diagnosis by three-dimensional facial imaging. Genetics in Medicine, 2020, 22, 1682-1693.	1.1	47
50	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. American Journal of Human Genetics, 2017, 101, 139-148.	2.6	45
51	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	1.4	45
52	Clinical genetics and the Hutterite population: A review of Mendelian disorders. American Journal of Medical Genetics, Part A, 2008, 146A, 1088-1098.	0.7	44
53	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
54	PISDis a mitochondrial disease gene causing skeletal dysplasia, cataracts, and white matter changes. Life Science Alliance, 2019, 2, e201900353.	1.3	41

#	Article	IF	Citations
55	Copy-number variations are enriched for neurodevelopmental genes in children with developmental coordination disorder. Journal of Medical Genetics, 2016, 53, 812-819.	1.5	40
56	Unique disease heritage of the Dutchâ€German Mennonite population. American Journal of Medical Genetics, Part A, 2008, 146A, 1072-1087.	0.7	38
57	The R941L mutation in MYH14 disrupts mitochondrial fission and associates with peripheral neuropathy. EBioMedicine, 2019, 45, 379-392.	2.7	37
58	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. American Journal of Human Genetics, 2015, 97, 862-868.	2.6	36
59	Expansion of phenotype and genotypic data in CRB2-related syndrome. European Journal of Human Genetics, 2016, 24, 1436-1444.	1.4	36
60	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	0.7	34
61	Matching Two Independent Cohorts ValidatesDPH1as a Gene Responsible for Autosomal Recessive Intellectual Disability with Short Stature, Craniofacial, and Ectodermal Anomalies. Human Mutation, 2015, 36, 1015-1019.	1.1	32
62	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogryposis. European Journal of Human Genetics, 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. European Journal of Human Genetics, 2019, 27, 738-746.	1.4	32
64	The value of diagnostic testing for parents of children with rare genetic diseases. Genetics in Medicine, 2019, 21, 2798-2806.	1.1	31
65	X-chromosome inactivation is mostly random in placental tissues of female monozygotic twins and triplets. , 1996, 61, 209-215.		30
66	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
67	Neuropathologic Features of Pontocerebellar Hypoplasia Type 6. Journal of Neuropathology and Experimental Neurology, 2014, 73, 1009-1025.	0.9	28
68	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	5.8	28
69	When One Diagnosis Is Not Enough. New England Journal of Medicine, 2017, 376, 83-85.	13.9	27
70	Phenotypic spectrum of Auâ€“Kline syndrome: a report of six new cases and review of the literature. European Journal of Human Genetics, 2018, 26, 1272-1281.	1.4	26
71	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	1.8	26
72	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	1.1	26

#	Article	IF	Citations
73	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
74	Refinement of the critical region of 1q41q42 microdeletion syndrome identifies FBXO28 as a candidate causative gene for intellectual disability and seizures. American Journal of Medical Genetics, Part A, 2014, 164, 441-448.	0.7	24
75	Homozygous Lamin A/C familial lipodystrophy R482Q mutation in autosomal recessive Emery Dreifuss muscular dystrophy. Neuromuscular Disorders, 2013, 23, 265-268.	0.3	23
76	Biallelic sequence variants in INTS1 in patients with developmental delays, cataracts, and craniofacial anomalies. European Journal of Human Genetics, 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. American Journal of Medical Genetics, Part A, 2006, 140A, 1375-1383.	0.7	22
78	SHORT syndrome due to a novel de novo mutation in PRKCE (Protein Kinase CÉ›) impairing TORC2-dependent AKT activation. Human Molecular Genetics, 2017, 26, 3713-3721.	1.4	22
79	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	1.0	21
80	Meckel syndrome in the Hutterite population is actually a Joubert-related cerebello-oculo-renal syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1715-1725.	0.7	20
81	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	1.7	20
82	Adult MTM1-related myopathy carriers. Neurology, 2019, 93, e1535-e1542.	1.5	18
83	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. American Journal of Human Genetics, 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. American Journal of Medical Genetics Part A, 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. American Journal of Human Genetics, 2018, 103, 786-793.	2.6	17
86	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
87	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17
88	De novo exon 1 missense mutations of SKI and Shprintzenâ€Goldberg syndrome: Two new cases and a clinical review. American Journal of Medical Genetics, Part A, 2014, 164, 676-684.	0.7	16
89	Nucleocytoplasmic transport of the RNA-binding protein CELF2 regulates neural stem cell fates. Cell Reports, 2021, 35, 109226.	2.9	16
90	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

#	Article	IF	Citations
91	A novel mutation in two Hmong families broadens the range of STRA6â€related malformations to include contractures and camptodactyly. American Journal of Medical Genetics, Part A, 2016, 170, 11-18.	0.7	15
92	Expansion of the <scp>GLE1</scp>â€associated arthrogryposis multiplex congenita clinical spectrum. Clinical Genetics, 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. Genetics in Medicine, 2022, 24, 100-108.	1.1	15
95	A shared founder mutation underlies restrictive dermopathy in Old Colony (Dutchâ€German) Mennonite and Hutterite patients in North America. American Journal of Medical Genetics, Part A, 2012, 158A, 1229-1232.	0.7	14
96	A novel NDUFS4 frameshift mutation causes Leigh disease in the Hutterite population. American Journal of Medical Genetics, Part A, 2017, 173, 596-600.	0.7	14
97	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	2.6	14
98	A rational approach to the child with mental retardation for the paediatrician. Paediatrics and Child Health, 2003, 8, 345-356.	0.3	13
99	Unsolved recognizable patterns of human malformation: Challenges and opportunities. , 2018, 178, 382-386.		13
100	Is PNPT1â€related hearing loss ever nonâ€syndromic? Whole exome sequencing of adult siblings expands the natural history of PNPT1â€related disorders. American Journal of Medical Genetics, Part A, 2018, 176, 2487-2493.	0.7	13
101	A locus for Bowen-Conradi syndrome maps to chromosome region 12p13.3. American Journal of Medical Genetics, Part A, 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. American Journal of Medical Genetics, Part A, 2010, 152A, 1349-1356.	0.7	12
103	The de novo CACNA1A pathogenic variant Y1384C associated with hemiplegic migraine, early onset cerebellar atrophy and developmental delay leads to a loss of Cav2.1 channel function. Molecular Brain, 2021, 14, 27.	1.3	12
104	When to think outside the autozygome: Best practices for exome sequencing in â€œconsanguineousâ€• families. Clinical Genetics, 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. Prenatal Diagnosis, 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 WDR35. American Journal of Medical Genetics, Part A, 2016, 170, 760-765.	0.7	10
107	Overlapping phenotypes between SHORT and Noonan syndromes in patients with PTPN11 pathogenic variants. Clinical Genetics, 2020, 98, 10-18.	1.0	9
108	Intracerebral hemorrhage in a young man. Cmaj, 2011, 183, E61-E64.	0.9	8

#	Article	IF	Citations
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 TRPM3 p.(<scp>Val837Met</scp>) 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 28â€yearâ€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 Cardioâ€“Facioâ€“Cutaneous Syndrome. The American Journal of Pediatric Hematology/oncology, 2000, 22, 546.	1.3	7
116	<scp>CHRNB1</scp>â€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 <scp>DNA</scp> 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 Fryns-like phenotype. Prenatal Diagnosis, 2017, 37, 602-610.	1.1	5
120	De novo variants in MPP5 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 â€˜Louâ€™ 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 carriersâ€”Authorsâ€™ reply. EBioMedicine, 2019, 49, 25.	2.7	2

#	Article	IF	Citations
127	Next-Generation Sequencing Using a Cardiac Gene Panel in Prenatally Diagnosed Cardiac Anomalies. Journal of Obstetrics and Gynaecology Canada, 2018, 40, 1417-1423.	0.3	2
128	Next Generation Diagnostics for Rare Neurological Diseases: The Future is Here. Canadian Journal of Neurological Sciences, 2014, 41, 299-300.	0.3	1
129	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
130	Molecular genetic testing and genetic counseling. Handbook of Clinical Neurology / 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. American Journal of Medical Genetics, Part A, 2008, 146A, 1067-1071.	0.7	0
132	Response to correspondence of NDUFS4â€related Leigh syndrome in Hutterites. American Journal of Medical Genetics, Part A, 2017, 173, 1452-1452.	0.7	0
133	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
134	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	2.6	0
135	Re: 3C (Ritscher-Schinzel) syndrome: the importance of ruling out a terminal 6p deletion. Clinical Dysmorphology, 2005, 14, 209-210.	0.1	0