

# GeneMatcher: A Matching Tool for Connecting Investigative Gene

Human Mutation

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Citation Report

#	ARTICLE	IF	CITATIONS
1	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. <i>Human Mutation</i> , 2015, 36, 931-940.	1.1	107
2	The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. <i>Human Mutation</i> , 2015, 36, 922-927.	1.1	50
3	Use of Model Organism and Disease Databases to Support Matchmaking for Human Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 979-984.	1.1	36
4	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 915-921.	1.1	390
5	Human genotypeâ€™ phenotype databases: aims, challenges and opportunities. <i>Nature Reviews Genetics</i> , 2015, 16, 702-715.	7.7	100
6	Exaptation of Bornavirus-Like Nucleoprotein Elements in Afrotherians. <i>PLoS Pathogens</i> , 2016, 12, e1005785.	2.1	26
7	Sequencing-based diagnostics for pediatric genetic diseases: progress and potential. <i>Expert Review of Molecular Diagnostics</i> , 2016, 16, 987-999.	1.5	29
8	De novo pathogenic variants in <i>CHAMP1</i> are associated with global developmental delay, intellectual disability, and dysmorphic facial features. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a000661.	0.5	31
9	New insights into the generation and role of de novo mutations in health and disease. <i>Genome Biology</i> , 2016, 17, 241.	3.8	339
10	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. <i>Genome Medicine</i> , 2016, 8, 106.	3.6	43
11	Mutations in <i>HIVEP2</i> are associated with developmental delay, intellectual disability, and dysmorphic features. <i>Neurogenetics</i> , 2016, 17, 159-164.	0.7	31
12	Clinical genomics: from a truly personal genome viewpoint. <i>Human Genetics</i> , 2016, 135, 591-601.	1.8	15
13	Novel bioinformatic developments for exome sequencing. <i>Human Genetics</i> , 2016, 135, 603-614.	1.8	37
14	Discovery of mutations for Mendelian disorders. <i>Human Genetics</i> , 2016, 135, 615-623.	1.8	53
15	A Clinicianâ€™s perspective on clinical exome sequencing. <i>Human Genetics</i> , 2016, 135, 643-654.	1.8	33
16	De Novo Truncating Variants in <i>ASXL2</i> Are Associated with a Unique and Recognizable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 991-999.	2.6	68
17	Biallelic Mutations in <i>TBCD</i> , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 99, 962-973.	2.6	66
18	<i>ARCN1</i> Mutations Cause a Recognizable Craniofacial Syndrome Due to <i>COPI</i> -Mediated Transport Defects. <i>American Journal of Human Genetics</i> , 2016, 99, 451-459.	2.6	65

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19	A <i>de novo</i> frameshift in <i>HNRNPK</i> causing a Kabuki-like syndrome with nodular heterotopia. <i>Clinical Genetics</i> , 2016, 90, 258-262.	1.0	33
20	Biallelic Variants in <i>UBA5</i> Link Dysfunctional <i>UBA5</i> -Ubiquitin-like Modifier Pathway to Severe Infantile-Onset Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 99, 683-694.	2.6	72
21	De novo <i>FGF12</i> mutation in 2 patients with neonatal-onset epilepsy. <i>Neurology: Genetics</i> , 2016, 2, e120.	0.9	29
22	Recurrent De Novo and Biallelic Variation of <i>ATAD3A</i> , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	2.6	146
23	De Novo Mutations in <i>CHD4</i> , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. <i>American Journal of Human Genetics</i> , 2016, 99, 934-941.	2.6	111
24	<i>MECR</i> Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. <i>American Journal of Human Genetics</i> , 2016, 99, 1229-1244.	2.6	91
25	Missense-depleted regions in population exomes implicate ras superfamily nucleotide-binding protein alteration in patients with brain malformation. <i>Npj Genomic Medicine</i> , 2016, 1, .	1.7	41
26	Truncating de novo mutations in the Krüppel-type zinc-finger gene <i>ZNF148</i> in patients with corpus callosum defects, developmental delay, short stature, and dysmorphisms. <i>Genome Medicine</i> , 2016, 8, 131.	3.6	24
27	Explorations to improve the completeness of exome sequencing. <i>BMC Medical Genomics</i> , 2016, 9, 56.	0.7	9
28	Gene discovery for Mendelian conditions via social networking: de novo variants in <i>KDM1A</i> cause developmental delay and distinctive facial features. <i>Genetics in Medicine</i> , 2016, 18, 788-795.	1.1	88
29	Monoallelic and Biallelic Variants in <i>EMC1</i> Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 562-570.	2.6	66
30	Autosomal recessive mutations in <i>THOC6</i> cause intellectual disability: syndrome delineation requiring forward and reverse phenotyping. <i>Clinical Genetics</i> , 2017, 91, 92-99.	1.0	28
31	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. <i>Human Mutation</i> , 2017, 38, 594-599.	1.1	3
32	A Recurrent De Novo Variant in <i>NACC1</i> Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. <i>American Journal of Human Genetics</i> , 2017, 100, 343-351.	2.6	35
33	De Novo Disruption of the Proteasome Regulatory Subunit <i>PSMD12</i> Causes a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 100, 352-363.	2.6	86
34	Mutations in <i>EXTL3</i> Cause Neuro-immuno-skeletal Dysplasia Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 281-296.	2.6	59
35	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase ( <i>TRIT1</i> ) gene. <i>Human Mutation</i> , 2017, 38, 511-516.	1.1	39
36	Mutations in <i>INPP5K</i> Cause a Form of Congenital Muscular Dystrophy Overlapping Marinesco-Sjögren Syndrome and Dystroglycanopathy. <i>American Journal of Human Genetics</i> , 2017, 100, 537-545.	2.6	67

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37	<i>STAG1</i> mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. <i>Journal of Medical Genetics</i> , 2017, 54, 479-488.	1.5	35
38	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , 2017, 100, 695-705.	2.6	305
39	“Matching” consent to purpose: The example of the Matchmaker Exchange. <i>Human Mutation</i> , 2017, 38, 1281-1285.	1.1	13
40	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. <i>European Journal of Human Genetics</i> , 2017, 25, 946-951.	1.4	33
41	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. <i>Brain</i> , 2017, 140, 2093-2103.	3.7	31
42	The role of genetic testing in epilepsy diagnosis and management. <i>Expert Review of Molecular Diagnostics</i> , 2017, 17, 739-750.	1.5	71
43	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 100, 907-925.	2.6	125
44	Genomic diagnosis for children with intellectual disability and/or developmental delay. <i>Genome Medicine</i> , 2017, 9, 43.	3.6	188
45	A RaDiCAL gene hunt. <i>Journal of Taibah University Medical Sciences</i> , 2017, 12, 194-198.	0.5	0
46	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 100, 676-688.	2.6	54
47	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	3.6	184
48	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 650-658.	2.6	56
49	A systematic review of genetic syndromes with obesity. <i>Obesity Reviews</i> , 2017, 18, 603-634.	3.1	138
50	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. <i>Human Genetics</i> , 2017, 136, 463-479.	1.8	66
51	Disruption of the ATXN1“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. <i>Nature Genetics</i> , 2017, 49, 527-536.	9.4	113
52	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 666-675.	2.6	22
53	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2017, 100, 117-127.	2.6	62
54	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. <i>Neuron</i> , 2017, 93, 115-131.	3.8	95

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55	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	2.6	72
56	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	2.6	63
57	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. American Journal of Human Genetics, 2017, 101, 824-832.	2.6	36
58	Biallelic MCM3AP mutations cause Charcot-Marie-Tooth neuropathy with variable clinical presentation. Brain, 2017, 140, e65-e65.	3.7	13
59	Survival beyond the perinatal period expands the phenotypes caused by mutations in <i>GLE1</i> . American Journal of Medical Genetics, Part A, 2017, 173, 3098-3103.	0.7	10
60	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
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62	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
63	Activating de novo mutations in NFE2L2 encoding NRF2 cause a multisystem disorder. Nature Communications, 2017, 8, 818.	5.8	72
64	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	2.6	43
65	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	2.6	61
66	De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects. Human Molecular Genetics, 2017, 26, 4849-4860.	1.4	42
67	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellis-van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571.	1.4	50
68	<i>GABBR2</i> mutations determine phenotype in rett syndrome and epileptic encephalopathy. Annals of Neurology, 2017, 82, 466-478.	2.8	66
69	Loss-of-function variants in <i>NFIA</i> provide further support that <i>NFIA</i> is a critical gene in 1p32-p31 deletion syndrome: A four patient series. American Journal of Medical Genetics, Part A, 2017, 173, 3158-3164.	0.7	16
70	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	1.8	122
71	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	2.6	35
72	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	2.6	119

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74	Survival among children with "Lethal" congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene ( <i>GLDN</i> ). <i>Human Mutation</i> , 2017, 38, 1477-1484.	1.1	19
75	Heterozygous De Novo UBTF Gain-of-Function Variant Is Associated with Neurodegeneration in Childhood. <i>American Journal of Human Genetics</i> , 2017, 101, 267-273.	2.6	41
76	De Novo Mutations in YWHAG Cause Early-Onset Epilepsy. <i>American Journal of Human Genetics</i> , 2017, 101, 300-310.	2.6	65
77	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. <i>New England Journal of Medicine</i> , 2017, 377, 544-552.	13.9	177
78	Deriving genomic diagnoses without revealing patient genomes. <i>Science</i> , 2017, 357, 692-695.	6.0	110
79	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitochondrial Subunit and Leigh Syndrome. <i>American Journal of Human Genetics</i> , 2017, 101, 239-254.	2.6	83
80	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. <i>American Journal of Human Genetics</i> , 2017, 101, 985-994.	2.6	44
81	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures" American Journal of Human Genetics, 2017, 101, 815-823.	2.6	37
82	Heterozygous variants in <i>ACTL6A</i> , encoding a component of the BAF complex, are associated with intellectual disability. <i>Human Mutation</i> , 2017, 38, 1365-1371.	1.1	27
83	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
84	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. <i>American Journal of Human Genetics</i> , 2017, 101, 149-156.	2.6	44
85	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2017, 101, 65-74.	2.6	99
86	Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. <i>Genetics in Medicine</i> , 2017, 19, 224-235.	1.1	47
87	Further evidence that <i>de novo</i> missense and truncating variants in <i>ZBTB18</i> cause intellectual disability with variable features. <i>Clinical Genetics</i> , 2017, 91, 697-707.	1.0	29
88	A recurrent <i>de novo</i> mutation in TMEM106B causes hypomyelinating leukodystrophy. <i>Brain</i> , 2017, 140, 3105-3111.	3.7	64
89	MYT1L mutations cause intellectual disability and variable obesity by dysregulating gene expression and development of the neuroendocrine hypothalamus. <i>PLoS Genetics</i> , 2017, 13, e1006957.	1.5	60
90	Computer face-matching technology using two-dimensional photographs accurately matches the facial gestalt of unrelated individuals with the same syndromic form of intellectual disability. <i>BMC Biotechnology</i> , 2017, 17, 90.	1.7	37

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92	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. <i>Cell</i> , 2018, 172, 924-936.e11.	13.5	103
93	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. <i>American Journal of Human Genetics</i> , 2018, 102, 995-1007.	2.6	49
94	Myonuclear Positioning and Aneurysms Are LINCâ€™d by Ariadne. <i>Developmental Cell</i> , 2018, 45, 149-150.	3.1	1
95	Ari-1 Regulates Myonuclear Organization Together with Parkin and Is Associated with Aortic Aneurysms. <i>Developmental Cell</i> , 2018, 45, 226-244.e8.	3.1	46
96	Systematic reanalysis of genomic data improves quality of variant interpretation. <i>Clinical Genetics</i> , 2018, 94, 174-178.	1.0	30
97	Mutations in <i>VPS13D</i> lead to a new recessive ataxia with spasticity and mitochondrial defects. <i>Annals of Neurology</i> , 2018, 83, 1075-1088.	2.8	122
98	The functional genomics laboratory: functional validation of genetic variants. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 297-307.	1.7	48
99	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 102, 309-320.	2.6	138
100	Exome and genome sequencing in reproductive medicine. <i>Fertility and Sterility</i> , 2018, 109, 213-220.	0.5	22
101	Paediatric genomics: diagnosing rare disease in children. <i>Nature Reviews Genetics</i> , 2018, 19, 253-268.	7.7	369
102	De novo variants in <i>CDK13</i> associated with syndromic ID/DD: Molecular and clinical delineation of 15 individuals and a further review. <i>Clinical Genetics</i> , 2018, 93, 1000-1007.	1.0	20
103	Biallelic variants in KIF14 cause intellectual disability with microcephaly. <i>European Journal of Human Genetics</i> , 2018, 26, 330-339.	1.4	52
104	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 468-479.	2.6	63
105	Biallelic CHP1 mutation causes human autosomal recessive ataxia by impairing NHE1 function. <i>Neurology: Genetics</i> , 2018, 4, e209.	0.9	23
106	De novo variants in <i>KLF7</i> are a potential novel cause of developmental delay/intellectual disability, neuromuscular and psychiatric symptoms. <i>Clinical Genetics</i> , 2018, 93, 1030-1038.	1.0	9
107	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 27-43.	2.6	88
108	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in <i>Drosophila</i> . <i>American Journal of Human Genetics</i> , 2018, 102, 44-57.	2.6	49

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109	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. <i>Nature Neuroscience</i> , 2018, 21, 207-217.	7.1	30
110	Toward clinical and molecular understanding of pathogenic variants in the <i>ZBTB18</i> gene. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 393-400.	0.6	22
111	De novo mutations in the <i>SET</i> nuclear proto-oncogene, encoding a component of the inhibitor of histone acetyltransferases (INHAT) complex in patients with nonsyndromic intellectual disability. <i>Human Mutation</i> , 2018, 39, 1014-1023.	1.1	18
112	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	2.6	51
113	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	2.6	59
114	Mutations in PMPCB Encoding the Catalytic Subunit of the Mitochondrial Presequence Protease Cause Neurodegeneration in Early Childhood. <i>American Journal of Human Genetics</i> , 2018, 102, 557-573.	2.6	69
115	Mutations in the mitochondrial ribosomal protein MRPS22 lead to primary ovarian insufficiency. <i>Human Molecular Genetics</i> , 2018, 27, 1913-1926.	1.4	39
116	Whole-exome sequencing is a valuable diagnostic tool for inherited peripheral neuropathies: Outcomes from a cohort of 50 families. <i>Clinical Genetics</i> , 2018, 93, 301-309.	1.0	48
117	Clinical and genetic heterogeneity in familial steroid-sensitive nephrotic syndrome. <i>Pediatric Nephrology</i> , 2018, 33, 473-483.	0.9	34
118	Missense variants in the chromatin remodeler <i>CHD1</i> are associated with neurodevelopmental disability. <i>Journal of Medical Genetics</i> , 2018, 55, 561-566.	1.5	49
119	Homozygosity for a nonsense variant in AIMP2 is associated with a progressive neurodevelopmental disorder with microcephaly, seizures, and spastic quadriplegia. <i>Journal of Human Genetics</i> , 2018, 63, 19-25.	1.1	26
120	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. <i>Neuron</i> , 2018, 100, 1354-1368.e5.	3.8	35
121	Loss of tubulin deglutamylase <i>CCP1</i> causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	3.5	86
122	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
123	Alphabet Soup: Recurrent De Novo Mutations in Novel Genes Causing Developmental and Epileptic Encephalopathies. <i>Epilepsy Currents</i> , 2018, 18, 125-127.	0.4	1
124	Diagnosing rare diseases after the exome. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003392.	0.5	48
125	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 948-967.	2.6	18
126	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. <i>PLoS Genetics</i> , 2018, 14, e1007671.	1.5	16



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127	Further delineation of the clinical spectrum of de novo <i>TRIM8</i> truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478.	0.7	19
128	<i>matchbox</i> : An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	1.1	20
129	Bi-allelic Mutations in <i>NDUFA6</i> Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	2.6	41
130	<i>NBEA</i> : Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	2.8	44
131	Next-Generation Sequencing to Diagnose Suspected Genetic Disorders. New England Journal of Medicine, 2018, 379, 1353-1362.	13.9	181
132	Mutations in <i>KCNK4</i> that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	2.6	73
133	ClinGen's GenomeConnect registry enables patient-centered data sharing. Human Mutation, 2018, 39, 1668-1676.	1.1	25
134	Bi-allelic <i>CCDC47</i> Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. American Journal of Human Genetics, 2018, 103, 794-807.	2.6	18
135	On the verge of diagnosis: Detection, reporting, and investigation of de novo variants in novel genes identified by clinical sequencing. Human Mutation, 2018, 39, 1505-1516.	1.1	9
136	De Novo Pathogenic Variants in <i>CACNA1E</i> Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	2.6	87
137	Missense Mutations of the Pro65 Residue of <i>PCGF2</i> Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American Journal of Human Genetics, 2018, 103, 786-793.	2.6	17
138	<i>NFIB</i> Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	2.6	40
139	Identification of Inherited Retinal Disease-Associated Genetic Variants in 11 Candidate Genes. Genes, 2018, 9, 21.	1.0	20
140	De novo variant in <i>KIF26B</i> is associated with pontocerebellar hypoplasia with infantile spinal muscular atrophy. American Journal of Medical Genetics, Part A, 2018, 176, 2623-2629.	0.7	19
141	Variable cardiovascular phenotypes associated with <i>SMAD2</i> pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	1.1	23
142	<i>SLC35A2</i> -related congenital disorder of glycosylation: Defining the phenotype. European Journal of Paediatric Neurology, 2018, 22, 1095-1102.	0.7	27
143	Visual impairment and progressive phthisis bulbi caused by recessive pathogenic variant in <i>MARK3</i> . Human Molecular Genetics, 2018, 27, 2703-2711.	1.4	21
144	A homozygous <i>KAT2B</i> variant modulates the clinical phenotype of <i>ADD3</i> deficiency in humans and flies. PLoS Genetics, 2018, 14, e1007386.	1.5	17

#	ARTICLE	IF	CITATIONS
145	Collaborative science unites researchers and a novel spastic ataxia gene. <i>Annals of Neurology</i> , 2018, 83, 1072-1074.	2.8	4
146	What's new in pontocerebellar hypoplasia? An update on genes and subtypes. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 92.	1.2	101
147	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018, 103, 305-316.	2.6	48
148	IRF2BPL Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	2.6	69
149	Haploinsufficiency of <i>CUX1</i> Causes Nonsyndromic Global Developmental Delay With Possible Catch-up Development. <i>Annals of Neurology</i> , 2018, 84, 200-207.	2.8	23
150	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. <i>American Journal of Human Genetics</i> , 2018, 103, 221-231.	2.6	65
151	Special Issue Introduction: Inherited Retinal Disease: Novel Candidate Genes, Genotype-Phenotype Correlations, and Inheritance Models. <i>Genes</i> , 2018, 9, 215.	1.0	58
152	Registered access: authorizing data access. <i>European Journal of Human Genetics</i> , 2018, 26, 1721-1731.	1.4	33
153	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. <i>Brain</i> , 2018, 141, 2299-2311.	3.7	81
154	Recent Developments in Using <i>Drosophila</i> as a Model for Human Genetic Disease. <i>International Journal of Molecular Sciences</i> , 2018, 19, 2041.	1.8	18
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156	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. <i>Human Genetics</i> , 2018, 137, 375-388.	1.8	46
157	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. <i>Human Molecular Genetics</i> , 2018, 27, 2454-2465.	1.4	54
158	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2348-2361.	3.0	147
159	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. <i>American Journal of Human Genetics</i> , 2018, 103, 431-439.	2.6	62
160	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogyposis. <i>European Journal of Human Genetics</i> , 2018, 26, 1752-1758.	1.4	32
161	Rapid Paediatric Sequencing (RaPS): comprehensive real-life workflow for rapid diagnosis of critically ill children. <i>Journal of Medical Genetics</i> , 2018, 55, 721-728.	1.5	98
162	Bi-allelic mutations in <i>TRAPPC2L</i> result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. <i>Journal of Medical Genetics</i> , 2018, 55, 753-764.	1.5	39

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164	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. <i>Brain</i> , 2018, 141, 1998-2013.	3.7	67
165	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	1.1	60
166	Mouse models as a tool for discovering new neurological diseases. <i>Neurobiology of Learning and Memory</i> , 2019, 165, 106902.	1.0	17
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169	Biallelic and <i>De Novo</i> Variants in <i>DONSON</i> Reveal a Clinical Spectrum of Cell Cycleopathies with Microcephaly, Dwarfism and Skeletal Abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2056-2066.	0.7	15
170	<i>MAGEL2</i> -related disorders: A study and case series. <i>Clinical Genetics</i> , 2019, 96, 493-505.	1.0	26
172	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. <i>American Journal of Human Genetics</i> , 2019, 105, 640-657.	2.6	31
173	Rare De Novo Missense Variants in RNA Helicase DDX6 Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. <i>American Journal of Human Genetics</i> , 2019, 105, 509-525.	2.6	50
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175	Expanding phenotype with severe midline brain anomalies and missense variant supports a causal role for <i>FOXA2</i> in 20p11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1783-1790.	0.7	10
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179	Cohesin complex-associated holoprosencephaly. <i>Brain</i> , 2019, 142, 2631-2643.	3.7	43
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301	Phenotype and mutation expansion of the PTPN23 associated disorder characterized by neurodevelopmental delay and structural brain abnormalities. European Journal of Human Genetics, 2020, 28, 76-87.	1.4	21
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309	Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. <i>Brain</i> , 2020, 143, 94-111.	3.7	18
310	The undiagnosed diseases program: Approach to diagnosis. <i>Translational Science of Rare Diseases</i> , 2020, 4, 179-188.	1.6	9
311	CSCALNACT1â€œcongenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. <i>Human Mutation</i> , 2020, 41, 655-667.	1.1	15
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318	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. <i>Genetics in Medicine</i> , 2020, 22, 736-744.	1.1	83
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360	Molecular testing for the study of non-syndromic hearing loss. Hearing, Balance and Communication, 2020, 18, 270-277.	0.1	5
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364	De novo missense variants in LMBRD2 are associated with developmental and motor delays, brain structure abnormalities and dysmorphic features. <i>Journal of Medical Genetics</i> , 2020, 58, jmedgenet-2020-107137.	1.5	3
365	<i>CIC de novo</i> loss of function variants contribute to cerebral folate deficiency by downregulating <i>FOLR1</i> expression. <i>Journal of Medical Genetics</i> , 2021, 58, 484-494.	1.5	12
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376	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020, 583, 90-95.	13.7	148
377	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. <i>Genetics in Medicine</i> , 2020, 22, 1215-1226.	1.1	22
378	Genomic sequencing highlights the diverse molecular causes of Perrault syndrome: a peroxisomal disorder (PEX6), metabolic disorders (CLPP, GGPS1), and mtDNA maintenance/translation disorders (LARS2, TFAM). <i>Human Genetics</i> , 2020, 139, 1325-1343.	1.8	21
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382	Mutations in the Kinesin-2 Motor <i>KIF3B</i> Cause an Autosomal-Dominant Ciliopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 893-904.	2.6	29
383	<i>De novo</i> variants in <i>SUPT16H</i> cause neurodevelopmental disorders associated with corpus callosum abnormalities. <i>Journal of Medical Genetics</i> , 2020, 57, 461-465.	1.5	7
384	Childhood rare lung disease in the 21st century: omics technology advances accelerating discovery. <i>Pediatric Pulmonology</i> , 2020, 55, 1828-1837.	1.0	8
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387	Autozygosity-driven genetic diagnosis in consanguineous families from Italy and the Greater Middle East. <i>Human Genetics</i> , 2020, 139, 1429-1441.	1.8	8
388	Biallelic variants in the RNA exosome gene <i>EXOSC5</i> are associated with developmental delays, short stature, cerebellar hypoplasia and motor weakness. <i>Human Molecular Genetics</i> , 2020, 29, 2218-2239.	1.4	19
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393	<i>De novo</i> <i>EIF2AK1</i> and <i>EIF2AK2</i> Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	2.6	37
394	Loss- or Gain-of-Function Mutations in <i>ACOX1</i> Cause Axonal Loss via Different Mechanisms. <i>Neuron</i> , 2020, 106, 589-606.e6.	3.8	71
395	Biallelic variants in <i>PSMB1</i> encoding the proteasome subunit $\beta^6$ cause impairment of proteasome function, microcephaly, intellectual disability, developmental delay and short stature. <i>Human Molecular Genetics</i> , 2020, 29, 1132-1143.	1.4	30
396	<i>De Novo</i> Frameshift Variants in the Neuronal Splicing Factor <i>NOVA2</i> Result in a Common C-Terminal Extension and Cause a Severe Form of Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2020, 106, 438-452.	2.6	17
397	A 2020 View on the Genetics of Developmental and Epileptic Encephalopathies. <i>Epilepsy Currents</i> , 2020, 20, 90-96.	0.4	39

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399	The frontiers of sequencing in undiagnosed neurodevelopmental diseases. <i>Current Opinion in Genetics and Development</i> , 2020, 65, 76-83.	1.5	6
400	<i>De novo</i> missense variants in the <sc><i>RAP1B</i></sc> gene identified in two patients with syndromic thrombocytopenia. <i>Clinical Genetics</i> , 2020, 98, 374-378.	1.0	5
401	<i>DLC5</i> variants are associated with multiple congenital anomalies including ciliopathy phenotypes. <i>Journal of Medical Genetics</i> , 2021, 58, 453-464.	1.5	10
402	<i>BAZ2B</i> haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. <i>Human Mutation</i> , 2020, 41, 921-925.	1.1	11
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411	<i>ZMYND11</i>-related syndromic intellectual disability: 16 patients delineating and expanding the phenotypic spectrum. <i>Human Mutation</i> , 2020, 41, 1042-1050.	1.1	20
412	Protein-elongating mutations in <i>MYH11</i> are implicated in a dominantly inherited smooth muscle dysmotility syndrome with severe esophageal, gastric, and intestinal disease. <i>Human Mutation</i> , 2020, 41, 973-982.	1.1	18
413	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. <i>European Journal of Human Genetics</i> , 2020, 28, 770-782.	1.4	27
414	Novel KIAA1033 / WASHC4 mutations in three patients with syndromic intellectual disability and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 792-797.	0.7	12
415	Jumonji domain containing 1C (JMJD1C) sequence variants in seven patients with autism spectrum disorder, intellectual disability and seizures. <i>European Journal of Medical Genetics</i> , 2020, 63, 103850.	0.7	3



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417	Bi-allelic Variants in <i>TKFC</i> Encoding Triokinase/FMN Cyclase Are Associated with Cataracts and Multisystem Disease. <i>American Journal of Human Genetics</i> , 2020, 106, 256-263.	2.6	16
418	Variants in <i>CAPZA2</i> , a member of an F-actin capping complex, cause intellectual disability and developmental delay. <i>Human Molecular Genetics</i> , 2020, 29, 1537-1546.	1.4	15
419	De novo variants in <i>CUL3</i> are associated with global developmental delays with or without infantile spasms. <i>Journal of Human Genetics</i> , 2020, 65, 727-734.	1.1	23
420	Oligosaccharyltransferase complex-related congenital disorders of glycosylation: A novel congenital disorder of glycosylation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1460-1465.	0.7	7
421	Bi-allelic <i>ADARB1</i> Variants Associated with Microcephaly, Intellectual Disability, and Seizures. <i>American Journal of Human Genetics</i> , 2020, 106, 467-483.	2.6	31
422	Bi-allelic Variants in the GPI Transamidase Subunit <i>PIGK</i> Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. <i>American Journal of Human Genetics</i> , 2020, 106, 484-495.	2.6	22
423	Bi-allelic Loss-of-Function Variants in <i>NUP188</i> Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. <i>American Journal of Human Genetics</i> , 2020, 106, 623-631.	2.6	18
424	A Cluster of Autism-Associated Variants on X-Linked <i>NLGN4X</i> Functionally Resemble <i>NLGN4Y</i> . <i>Neuron</i> , 2020, 106, 759-768.e7.	3.8	45
425	Pathogenic variants in <i>TNRC6B</i> cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. <i>Journal of Medical Genetics</i> , 2020, 57, 717-724.	1.5	14
426	Next-generation sequencing approaches and challenges in the diagnosis of developmental anomalies and intellectual disability. <i>Clinical Genetics</i> , 2020, 98, 433-444.	1.0	20
427	<i>HNRNP1</i> -related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. <i>Clinical Genetics</i> , 2020, 98, 91-98.	1.0	25
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429	<i>RSRC1</i> loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. <i>Brain</i> , 2020, 143, e31-e31.	3.7	6
430	Biallelic <i>GRM7</i> variants cause epilepsy, microcephaly, and cerebral atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 610-627.	1.7	15
431	Novel congenital disorder of <i>O</i> -linked glycosylation caused by <i>GALNT2</i> loss of function. <i>Brain</i> , 2020, 143, 1114-1126.	3.7	46
432	De novo variants in <i>SIAH1</i> , encoding an E3 ubiquitin ligase, are associated with developmental delay, hypotonia and dysmorphic features. <i>Journal of Medical Genetics</i> , 2021, 58, 205-212.	1.5	6
433	Impact of integrated translational research on clinical exome sequencing. <i>Genetics in Medicine</i> , 2021, 23, 498-507.	1.1	24

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435	Genetic testing in dementia – utility and clinical strategies. <i>Nature Reviews Neurology</i> , 2021, 17, 23-36.	4.9	26
436	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. <i>Journal of Medical Genetics</i> , 2021, 58, 400-413.	1.5	18
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438	De novo variants in MED12 cause X-linked syndromic neurodevelopmental disorders in 18 females. <i>Genetics in Medicine</i> , 2021, 23, 645-652.	1.1	18
439	Heterozygous loss of <i>WBP11</i> function causes multiple congenital defects in humans and mice. <i>Human Molecular Genetics</i> , 2021, 29, 3662-3678.	1.4	14
440	<i>FBXO28</i> causes developmental and epileptic encephalopathy with profound intellectual disability. <i>Epilepsia</i> , 2021, 62, e13-e21.	2.6	8
441	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. <i>Genetics in Medicine</i> , 2021, 23, 740-750.	1.1	25
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444	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. <i>European Journal of Human Genetics</i> , 2021, 29, 411-421.	1.4	13
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460	Biallelic <i>UBE4A</i> loss-of-function variants cause intellectual disability and global developmental delay. <i>Genetics in Medicine</i> , 2021, 23, 661-668.	1.1	2
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515	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	3.6	50
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519	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. <i>Human Genetics</i> , 2021, 140, 1061-1076.	1.8	4
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527	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 929-941.	2.6	15
528	A new syndrome of moyamoya disease, kidney dysplasia, aminotransferase elevation, and skin disease associated with <i>de novo</i> variants in <i>RNF213</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2168-2174.	0.7	8
529	Next-generation sequencing and the evolution of data sharing. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2633-2635.	0.7	4
530	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	1.8	18
531	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> associated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	1.1	18
532	PPFIA4 mutation: A second hit in POLG related disease?. <i>Epilepsy and Behavior Reports</i> , 2021, 16, 100455.	0.5	2
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537	Identification of missense <i>MAB21L1</i> variants in microphthalmia and aniridia. <i>Human Mutation</i> , 2021, 42, 877-890.	1.1	13
538	Biallelic VPS16 variants limit HOPS/CORVET levels and cause a mucopolysaccharidosis-like disease. <i>EMBO Molecular Medicine</i> , 2021, 13, e13376.	3.3	16
539	Deep-Phenotyping the Less Severe Spectrum of PIGT Deficiency and Linking the Gene to Myoclonic Atonic Seizures. <i>Frontiers in Genetics</i> , 2021, 12, 663643.	1.1	6
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541	Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. <i>Genetics in Medicine</i> , 2021, 23, 1738-1745.	1.1	7
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543	Biallelic variants in <i>HPDL</i> cause pure and complicated hereditary spastic paraplegia. <i>Brain</i> , 2021, 144, 1422-1434.	3.7	22
544	Syndromic neurodevelopmental disorder associated with de novo variants in <i>DDX23</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2863-2872.	0.7	8
545	Familial Autonomic Ganglionopathy Caused by Rare <i>CHRNA3</i> Genetic Variants. <i>Neurology</i> , 2021, 97, e145-e155.	1.5	12
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548	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. <i>Nature Communications</i> , 2021, 12, 2678.	5.8	26
549	Curation and expansion of Human Phenotype Ontology for defined groups of inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 369-378.	1.5	16
550	The contributions of careful clinical observations: A legacy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3202-3207.	0.7	1
551	Loss of function mutations in <i>GEMIN5</i> cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	5.8	28
552	<i>CSNK2B</i> : A broad spectrum of neurodevelopmental disability and epilepsy severity. <i>Epilepsia</i> , 2021, 62, e103-e109.	2.6	13
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564	Heterozygous <i>HMGB1</i> loss-of-function variants are associated with developmental delay and microcephaly. Clinical Genetics, 2021, 100, 386-395.	1.0	3
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569	NAPG mutation in family members with hereditary hemorrhagic telangiectasia in China. BMC Pulmonary Medicine, 2021, 21, 197.	0.8	2
570	Expanding the genetic landscape of oral-facial-digital syndrome with two novel genes. American Journal of Medical Genetics, Part A, 2021, 185, 2409-2416.	0.7	9
571	Proximal variants in <i>CCND2</i> associated with microcephaly, short stature, and developmental delay: A case series and review of inverse brain growth phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 2719-2738.	0.7	14
572	PRICKLE2 revisited—further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244.	1.4	5



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574	Biallelic truncating variants in <i>ATP9A</i> cause a novel neurodevelopmental disorder involving postnatal microcephaly and failure to thrive. <i>Journal of Medical Genetics</i> , 2022, 59, 662-668.	1.5	9
575	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2021, 23, 1912-1921.	1.1	5
576	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. <i>American Journal of Human Genetics</i> , 2021, 108, 1330-1341.	2.6	18
577	Identification of autosomal recessive nonsyndromic hearing impairment genes through the study of consanguineous and non-consanguineous families: past, present, and future. <i>Human Genetics</i> , 2022, 141, 413-430.	1.8	2
578	Human autoinflammatory disease reveals ELF4 as a transcriptional regulator of inflammation. <i>Nature Immunology</i> , 2021, 22, 1118-1126.	7.0	30
580	Pathogenic <i>MAST3</i> Variants in the <i>STK</i> Domain Are Associated with Epilepsy. <i>Annals of Neurology</i> , 2021, 90, 274-284.	2.8	7
581	<i>KCND2</i> variants associated with global developmental delay differentially impair Kv4.2 channel gating. <i>Human Molecular Genetics</i> , 2021, 30, 2300-2314.	1.4	12
582	Tenorio syndrome: Description of 14 novel cases and review of the clinical and molecular features. <i>Clinical Genetics</i> , 2021, 100, 405-411.	1.0	2
583	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. <i>Genetics in Medicine</i> , 2021, 23, 2138-2149.	1.1	11
584	Epileptic encephalopathy caused by <i>ARV1</i> deficiency: Refinement of the genotype-phenotype spectrum and functional impact on GPI-anchored proteins. <i>Clinical Genetics</i> , 2021, 100, 607-614.	1.0	6
585	<i>ZMYND11</i> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. <i>Clinical Genetics</i> , 2021, 100, 412-429.	1.0	5
586	Variants in LSM7 impair LSM complexes assembly, neurodevelopment in zebrafish and may be associated with an ultra-rare neurological disease. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100034.	1.0	3
587	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. <i>American Journal of Human Genetics</i> , 2021, 108, 1342-1349.	2.6	9
588	The genes of <i>OMIM</i> : A legacy of Victor McKusick. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3276-3283.	0.7	4
589	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. <i>Nature Genetics</i> , 2021, 53, 1006-1021.	9.4	44
590	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. <i>Brain Sciences</i> , 2021, 11, 931.	1.1	7
591	Missense NAA20 variants impairing the NatB protein N-terminal acetyltransferase cause autosomal recessive developmental delay, intellectual disability, and microcephaly. <i>Genetics in Medicine</i> , 2021, 23, 2213-2218.	1.1	11

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596	Biallelic <i>PI4KA</i> variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. <i>Brain</i> , 2021, 144, 2659-2669.	3.7	19
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