Lisenka E L M Vissers

List of Publications by Year
in descending order

144 papers	17,738 citations	19636 61 h-index	15716 125 g-index
163 all docs	163 docs citations	163 times ranked	23988 citing authors

#	Article	IF	Citations
1	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	13.9	1,367
2	Mutations in a new member of the chromodomain gene family cause CHARGE syndrome. Nature Genetics, 2004, 36, 955-957.	9.4	1,098
3	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	13.7	996
4	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	9.4	751
5	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
6	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	7.7	614
7	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
8	Diagnostic Genome Profiling in Mental Retardation. American Journal of Human Genetics, 2005, 77, 606-616.	2.6	514
9	Array-Based Comparative Genomic Hybridization for the Genomewide Detection of Submicroscopic Chromosomal Abnormalities. American Journal of Human Genetics, 2003, 73, 1261-1270.	2.6	423
10	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	9.4	418
11	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	7.1	407
12	CHARGE syndrome: the phenotypic spectrum of mutations in the CHD7 gene. Journal of Medical Genetics, 2005, 43, 306-314.	1.5	382
13	A recent bottleneck of Y chromosome diversity coincides with a global change in culture. Genome Research, 2015, 25, 459-466.	2.4	348
14	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
15	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	13.5	322
16	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	9.4	293
17	Parent-of-origin-specific signatures of de novo mutations. Nature Genetics, 2016, 48, 935-939.	9.4	266
18	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	9.4	236

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19	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
20	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	1.1	220
21	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	2.6	219
22	Post-zygotic Point Mutations Are an Underrecognized Source of De Novo Genomic Variation. American Journal of Human Genetics, 2015, 97, 67-74.	2.6	215
23	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. American Journal of Human Genetics, 2012, 91, 73-82.	2.6	214
24	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	9.4	194
25	Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2008, 45, 710-720.	1.5	191
26	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585.	9.4	191
27	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	9.4	175
28	Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1073-1081.	2.6	159
29	Mutations inDYNC1H1cause severe intellectual disability with neuronal migration defects. Journal of Medical Genetics, 2012, 49, 179-183.	1.5	151
30	A systematic review and standardized clinical validity assessment of male infertility genes. Human Reproduction, 2019, 34, 932-941.	0.4	144
31	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. Human Molecular Genetics, 2009, 18, 3579-3593.	1.4	143
32	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	1.1	143
33	Identification of disease genes by whole genome CGH arrays. Human Molecular Genetics, 2005, 14, R215-R223.	1.4	140
34	Recurrence and variability of germline EPCAM deletions in Lynch syndrome. Human Mutation, 2011, 32, 407-414.	1.1	137
35	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
36	Genomic microarrays in mental retardation: from copy number variation to gene, from research to diagnosis. Journal of Medical Genetics, 2010, 47, 289-297.	1.5	135

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37	Is the $1000 Genome as Near as We Think? A Cost Analysis of Next-Generation Sequencing. Clinical Chemistry, 2016, 62, 1458-1464.	1.5	126
38	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
39	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
40	Non-invasive prenatal diagnosis of fetal aneuploidies using massively parallel sequencing-by-ligation and evidence that cell-free fetal DNA in the maternal plasma originates from cytotrophoblastic cells. Expert Opinion on Biological Therapy, 2012, 12, S19-S26.	1.4	111
41	Dominant Î²-catenin mutations cause intellectual disability with recognizable syndromic features. Journal of Clinical Investigation, 2014, 124, 1468-1482.	3.9	110
42	Genotypeâ€“phenotype mapping of chromosome 18q deletions by highâ€resolution array CGH: An update of the phenotypic map. American Journal of Medical Genetics, Part A, 2007, 143A, 1858-1867.	0.7	106
43	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448.	1.1	105
44	Recurrent De Novo Mutations in PACS1 Cause Defective Cranial-Neural-Crest Migration and Define a Recognizable Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 1122-1127.	2.6	96
45	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	2.6	96
46	De novo copy number variants associated with intellectual disability have a paternal origin and age bias. Journal of Medical Genetics, 2011, 48, 776-778.	1.5	95
47	TRIOloss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. Human Molecular Genetics, 2016, 25, 892-902.	1.4	94
48	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	1.5	93
49	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. Human Genetics, 2015, 134, 97-109.	1.8	93
50	B56Î´-related protein phosphatase 2A dysfunction identified in patients with intellectual disability. Journal of Clinical Investigation, 2015, 125, 3051-3062.	3.9	91
51	Involvement of the kinesin family membersKIF4AandKIF5Cin intellectual disability and synaptic function. Journal of Medical Genetics, 2014, 51, 487-494.	1.5	90
52	Chondrodysplasia and Abnormal Joint Development Associated with Mutations in IMPAD1, Encoding the Golgi-Resident Nucleotide Phosphatase, gPAPP. American Journal of Human Genetics, 2011, 88, 608-615.	2.6	88
53	De Novo Mutations of the Gene Encoding the Histone Acetyltransferase KAT6B Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 290-294.	2.6	86
54	Mobster: accurate detection of mobile element insertions in next generation sequencing data. Genome Biology, 2014, 15, 488.	3.8	86

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55	Clinical and cytogenetic characterization of 13 Dutch patients with deletion 9p syndrome: Delineation of the critical region for a consensus phenotype. American Journal of Medical Genetics, Part A, 2008, 146A, 1430-1438.	0.7	85
56	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. American Journal of Human Genetics, 2017, 101, 478-484.	2.6	84
57	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	5.8	82
58	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American Journal of Human Genetics, 2016, 99, 711-719.	2.6	81
59	Heterozygous Mutations of FREM1 Are Associated with an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. PLoS Genetics, 2011, 7, e1002278.	1.5	80
60	Understanding the Psychosocial Effects of WES Test Results on Parents of Children with Rare Diseases. Journal of Genetic Counseling, 2016, 25, 1207-1214.	0.9	73
61	12p-Amplicon structure analysis in testicular germ cell tumors of adolescents and adults by array CGH. Oncogene, 2003, 22, 7695-7701.	2.6	72
62	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	2.6	65
63	GATAD2Bloss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth inDrosophila. Journal of Medical Genetics, 2013, 50, 507-514.	1.5	63
64	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. European Journal of Human Genetics, 2015, 23, 317-324.	1.4	61
65	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	2.6	59
66	Microdeletion and Microduplication Syndromes. Methods in Molecular Biology, 2012, 838, 29-75.	0.4	58
67	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	1.1	58
68	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	2.6	56
69	1 in 38 individuals at risk of a dominant medically actionable disease. European Journal of Human Genetics, 2019, 27, 325-330.	1.4	56
70	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
71	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	1.1	49
72	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49

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73	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
74	Wholeâ€exome sequencing detects somatic mutations of IDH1 in metaphyseal chondromatosis with <scp>D</scp>â€2â€hydroxyglutaric aciduria (MCâ€HGA). American Journal of Medical Genetics, Part A, 2011, 155, 2609-2616.	0.7	47
75	Variation of CNV distribution in five different ethnic populations. Cytogenetic and Genome Research, 2007, 118, 19-30.	0.6	46
76	Validation and application of a novel integrated genetic screening method to a cohort of 1,112 men with idiopathic azoospermia or severe oligozoospermia. Human Mutation, 2017, 38, 1592-1605.	1.1	45
77	Point mutations as a source of de novo genetic disease. Current Opinion in Genetics and Development, 2013, 23, 257-263.	1.5	44
78	Ovotestes and XY sex reversal in a female with an interstitial9q33.3-q34.1 deletion encompassingNR5A1 andLMX1B causing features of genitopatellar syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1071-1081.	0.7	43
79	Absence of Î±- and Î²-dystroglycan is associated with Walker-Warburg syndrome. Neurology, 2015, 84, 2177-2182.	1.5	40
80	De Novo Mutations Affecting the Catalytic CÎ± Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 104, 139-156.	2.6	39
81	Disruption of Teashirt Zinc Finger Homeobox 1 Is Associated with Congenital Aural Atresia in Humans. American Journal of Human Genetics, 2011, 89, 813-819.	2.6	38
82	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
83	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	5.8	38
84	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	1.1	37
85	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	2.6	37
86	Exome sequencing reveals novel causes as well as new candidate genes for human globozoospermia. Human Reproduction, 2020, 35, 240-252.	0.4	37
87	Chromosomal breakpoint mapping by arrayCGH using flow-sorted chromosomes. BioTechniques, 2003, 35, 1066-1070.	0.8	36
88	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	1.4	34
89	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252.	1.4	34
90	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	1.1	34

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91	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
92	De novo lossâ€ofâ€function mutations in Xâ€linked <scp>SMC1A</scp> cause severe <scp>ID</scp> and therapyâ€resistant epilepsy in females: expanding the phenotypic spectrum. Clinical Genetics, 2016, 90, 413-419.	1.0	32
93	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	1.4	32
94	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
95	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	2.6	31
96	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
97	Squalene Synthase Deficiency: Clinical, Biochemical, and Molecular Characterization of a Defect in Cholesterol Biosynthesis. American Journal of Human Genetics, 2018, 103, 125-130.	2.6	29
98	Recurrent Inversion Events at 17q21.31 Microdeletion Locus Are Linked to the <i>MAPT</i> H2 Haplotype. Cytogenetic and Genome Research, 2010, 129, 275-279.	0.6	27
99	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	2.6	27
100	Long-read trio sequencing of individuals with unsolved intellectual disability. European Journal of Human Genetics, 2021, 29, 637-648.	1.4	27
101	Complex chromosome 17p rearrangements associated with low-copy repeats in two patients with congenital anomalies. Human Genetics, 2007, 121, 697-709.	1.8	26
102	A compound heterozygous mutation in DPAGT1 results in a congenital disorder of glycosylation with a relatively mild phenotype. European Journal of Human Genetics, 2013, 21, 844-849.	1.4	25
103	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. PLoS ONE, 2014, 9, e112687.	1.1	23
104	A novel microdeletion, del(2)(q22.3q23.3) in a mentally retarded patient, detected by array-based comparative genomic hybridization. Clinical Genetics, 2004, 65, 429-432.	1.0	22
105	Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. American Journal of Human Genetics, 2012, 90, 599-613.	2.6	22
106	Diagnostic Exome Sequencing in Persons With Severe Intellectual Disability. Obstetrical and Gynecological Survey, 2013, 68, 191-193.	0.2	22
107	Evaluating a counselling strategy for diagnostic WES in paediatric neurology: an exploration of parents' information and communication needs. Clinical Genetics, 2016, 89, 244-250.	1.0	22
108	Systematic analysis of short tandem repeats in 38,095 exomes provides an additional diagnostic yield. Genetics in Medicine, 2021, 23, 1569-1573.	1.1	21

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109	How to proceed after â€œnegativeâ€•exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. Journal of Inherited Metabolic Disease, 2022, 45, 663-681.	1.7	20
110	Human disease genes website series: An international, open and dynamic library for upâ€toâ€date clinical information. American Journal of Medical Genetics, Part A, 2021, 185, 1039-1046.	0.7	19
111	Establishing the phenotypic spectrum of ZTTK syndrome by analysis of 52 individuals with variants in SON. European Journal of Human Genetics, 2022, 30, 271-281.	1.4	19
112	Two families with sibling recurrence of the 17q21.31 microdeletion syndrome due to low-grade mosaicism. European Journal of Human Genetics, 2012, 20, 729-733.	1.4	17
113	Standardized phenotyping enhances Mendelian disease gene identification. Nature Genetics, 2015, 47, 1222-1224.	9.4	17
114	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	1.4	17
115	SON haploinsufficiency causes impaired pre-mRNA splicing of CAKUT genes and heterogeneous renalÂphenotypes. Kidney International, 2019, 95, 1494-1504.	2.6	17
116	Reanalysis of exome negative patients with rare disease: a pragmatic workflow for diagnostic applications. Genome Medicine, 2022, 14, .	3.6	17
117	Reliable noninvasive prenatal testing by massively parallel sequencing of circulating cell-free DNA from maternal plasma processed up to 24h after venipuncture. Clinical Biochemistry, 2013, 46, 1783-1786.	0.8	15
118	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	1.4	15
119	Lessons learned from unsolicited findings in clinical exome sequencing of 16,482 individuals. European Journal of Human Genetics, 2022, 30, 170-177.	1.4	15
120	Characterization of the <scp>GABRB2</scp>â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2021, 89, 573-586.	2.8	14
121	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. Genomics Data, 2014, 2, 144-146.	1.3	13
122	Quantitative facial phenotyping for Koolen-de Vries and 22q11.2 deletion syndrome. European Journal of Human Genetics, 2021, 29, 1418-1423.	1.4	12
123	De novo mutations in children born after medical assisted reproduction. Human Reproduction, 2022, 37, 1360-1369.	0.4	12
124	Missense variants in ANKRD11 cause KBG syndrome by impairment of stability or transcriptional activity of the encoded protein. Genetics in Medicine, 2022, 24, 2051-2064.	1.1	12
125	An Emerging Female Phenotype with Lossâ€ofâ€Function Mutations in the Aristalessâ€ Related Homeodomain Transcription Factor ARX. Human Mutation, 2017, 38, 548-555.	1.1	10
126	Cell-based assay for ciliopathy patients to improve accurate diagnosis using ALPACA. European Journal of Human Genetics, 2021, 29, 1677-1689.	1.4	10

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127	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
128	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	2.6	8
129	FAIR Genomes metadata schema promoting Next Generation Sequencing data reuse in Dutch healthcare and research. Scientific Data, 2022, 9, 169.	2.4	8
130	DeNovoCNN: a deep learning approach to de novo variant calling in next generation sequencing data. Nucleic Acids Research, 2022, 50, e97-e97.	6.5	8
131	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	1.4	7
132	Congenital anomalies and genetic disorders in neonates and infants: a single-center observational cohort study. European Journal of Pediatrics, 2022, 181, 359-367.	1.3	7
133	Functional disruption of pyrimidine nucleoside transporter CNT1 results in a novel inborn error of metabolism with high excretion of uridine and cytidine. Journal of Inherited Metabolic Disease, 2019, 42, 494-500.	1.7	6
134	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. Genetics in Medicine, 2022, 24, 645-653.	1.1	6
135	Overrepresentation of genetic variation in the AnkyrinG interactome is related to a range of neurodevelopmental disorders. European Journal of Human Genetics, 2020, 28, 1726-1733.	1.4	4
136	Genetic convergence of developmental and epileptic encephalopathies and intellectual disability. Developmental Medicine and Child Neurology, 2021, 63, 1441-1447.	1.1	4
137	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. Human Genetics and Genomics Advances, 2021, 2, 100046.	1.0	4
138	Medical costs of children admitted to the neonatal intensive care unit: The role and possible economic impact of WES in early diagnosis. European Journal of Medical Genetics, 2022, 65, 104467.	0.7	3
139	Detecting fetal subchromosomal aberrations by MPS: an unexpected discrepancy between amniocyte DNA and ccffDNA. Prenatal Diagnosis, 2014, 34, 402-405.	1.1	2
140	A Next-Generation Framework: Deciding On The Role Of Costs In The Clinical Use Of Targeted Gene Panels, Exome And Genome Sequencing. Value in Health, 2015, 18, A352.	0.1	2
141	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. European Journal of Medical Genetics, 2022, 65, 104402.	0.7	2
142	Improved detection of CFTR variants by targeted next-generation sequencing in male infertility: a case series. Reproductive BioMedicine Online, 2019, 39, 963-968.	1.1	1
143	Economic evaluations of exome and genome sequencing in pediatric genetics: considerations towards a consensus strategy. Journal of Medical Economics, 2021, 24, 60-70.	1.0	1
144	Exome Sequencing of Late Recurrence T-Cell Acute Lymphoblastic Leukemia in Children Confirms Second Leukemia and Exposes Predisposition Candidate Genes. Blood, 2011, 118, 755-755.	0.6	0