Alessandra Renieri

List of Publications by Year
in descending order

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337	17,517	24978	22102
papers	citations	57 h-index	113 g-index

362	362	362	22591
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	1.8	12
2	Novel retinal finding in a patient with 4q12 deletion. Ophthalmic Genetics, 2022, 43, 120-122.	0.5	0
3	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis. Neurological Sciences, 2022, 43, 2849-2852.	0.9	0
4	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	1.8	22
5	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males. Autophagy, 2022, 18, 1662-1672.	4.3	25
6	The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516.	1.4	12
7	Predictive genetic testing for Motor neuron disease: time for a guideline?. European Journal of Human Genetics, 2022, 30, 635-636.	1.4	2
8	Development and Implementation of the AIDA International Registry for Patients With Still's Disease. Frontiers in Medicine, 2022, 9, 878797.	1.2	9
9	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	2.2	41
10	Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154.	2.2	49
11	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
12	Identification of a Novel SHANK2 Pathogenic Variant in a Patient with a Neurodevelopmental Disorder. Genes, 2022, 13, 688.	1.0	7
13	Understanding the new <scp>BRD4</scp>â€related syndrome: Clinical and genomic delineation with an international cohort study. Clinical Genetics, 2022, 102, 117-122.	1.0	3
14	Host genetic basis of COVID-19: from methodologies to genes. European Journal of Human Genetics, 2022, 30, 899-907.	1.4	13
15	Multiomic analysis reveals cell-type-specific molecular determinants of COVID-19 severity. Cell Systems, 2022, 13, 598-614.e6.	2.9	10
16	Carriers of ADAMTS13 Rare Variants Are at High Risk of Life-Threatening COVID-19. Viruses, 2022, 14, 1185.	1.5	1
17	Novel genes and sex differences in COVID-19 severity. Human Molecular Genetics, 2022, 31, 3789-3806.	1.4	38
18	Possible association of 16p11.2 copy number variation with altered lymphocyte and neutrophil counts. Npj Genomic Medicine, 2022, 7, .	1.7	3

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19	Pathogen-sugar interactions revealed by universal saturation transfer analysis. Science, 2022, 377, .	6.0	24
20	Geographical distribution of cystic fibrosis carriers as population genetic determinant of COVID-19 spread and fatality in 37 countries. Journal of Infection, 2022, 85, 318-321.	1.7	6
21	Nosological and Theranostic Approach to Vascular Malformation through cfDNA NGS Liquid Biopsy. Journal of Clinical Medicine, 2022, 11, 3740.	1.0	8
22	Digenic Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 1697-1706.	2.2	19
23	A pilot study of next generation sequencingâ€“liquid biopsy on cell-free DNA as a novel non-invasive diagnostic tool for Klippelâ€“Trenaunay syndrome. Vascular, 2021, 29, 85-91.	0.4	14
24	The effect of angiotensin-converting enzyme levels on COVID-19 susceptibility and severity: a Mendelian randomization study. International Journal of Epidemiology, 2021, 50, 75-86.	0.9	10
25	MET somatic activating mutations are responsible for lymphovenous malformation and can be identified using cell-free DNA next generation sequencing liquid biopsy. Journal of Vascular Surgery: Venous and Lymphatic Disorders, 2021, 9, 740-744.	0.9	7
26	CDKL5 mutations may mimic Pitt-Hopkins syndrome phenotype. European Journal of Medical Genetics, 2021, 64, 104102.	0.7	0
27	A new mutation in DNM2 gene in a large Italian family. Neurological Sciences, 2021, 42, 2509-2513.	0.9	1
28	<scp>IQSEC2</scp> disorder: A new disease entity or a Rett spectrum continuum?. Clinical Genetics, 2021, 99, 462-474.	1.0	11
29	Employing a systematic approach to biobanking and analyzing clinical and genetic data for advancing COVID-19 research. European Journal of Human Genetics, 2021, 29, 745-759.	1.4	35
30	Anakinra and canakinumab for patients with R92Q-associated autoinflammatory syndrome: a multicenter observational study from the AIDA Network. Therapeutic Advances in Musculoskeletal Disease, 2021, 13, 1759720X2110371.	1.2	1
31	Epilepsy in Nicolaidesâ€“Baraitser Syndrome: Review of Literature and Report of 25 Patients Focusing on Treatment Aspects. Neuropediatrics, 2021, 52, 109-122.	0.3	2
32	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	2.7	52
33	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males: findings from a nested case-control study. ELife, 2021, 10, .	2.8	145
34	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	1.0	39
35	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. European Journal of Human Genetics, 2021, 29, 1186-1197.	1.4	61
36	Solving unsolved rare neurological diseasesâ€”a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	1.4	4

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37	Drug survival of anakinra and canakinumab in monogenic autoinflammatory diseases: observational study from the International AIDA Registry. Rheumatology, 2021, 60, 5705-5712.	0.9	4
38	The phenomenon of multidrug resistance in glioblastomas. Hematology/ Oncology and Stem Cell Therapy, 2021, , .	0.6	10
39	In response to the letter to the editor by Soha Ghanian etÂal. re our publication â€œShorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European malesâ€• EBioMedicine, 2021, 68, 103426.	2.7	0
40	Severe COVID-19 in Hospitalized Carriers of Single CFTR Pathogenic Variants. Journal of Personalized Medicine, 2021, 11, 558.	1.1	16
41	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
42	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
43	C9orf72 Intermediate Repeats Confer Genetic Risk for Severe COVID-19 Pneumonia Independently of Age. International Journal of Molecular Sciences, 2021, 22, 6991.	1.8	12
44	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
45	Vitamin D and COVID-19 susceptibility and severity in the COVID-19 Host Genetics Initiative: A Mendelian randomization study. PLoS Medicine, 2021, 18, e1003605.	3.9	91
46	Clinical, molecular and glycophenotype insights in SLC39A8-CDG. Orphanet Journal of Rare Diseases, 2021, 16, 307.	1.2	4
47	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. Brain Sciences, 2021, 11, 936.	1.1	17
48	Biotechnological Agents for Patients With Tumor Necrosis Factor Receptor Associated Periodic Syndromeâ€”Therapeutic Outcome and Predictors of Response: Real-Life Data From the AIDA Network. Frontiers in Medicine, 2021, 8, 668173.	1.2	6
49	Mapping the human genetic architecture of COVID-19. Nature, 2021, 600, 472-477.	13.7	640
50	SELP Asp603Asn and severe thrombosis in COVID-19 males. Journal of Hematology and Oncology, 2021, 14, 123.	6.9	11
51	13q Deletion Syndrome Involving RB1: Characterization of a New Minimal Critical Region for Psychomotor Delay. Genes, 2021, 12, 1318.	1.0	2
52	Age-dependent impact of the major common genetic risk factor for COVID-19 on severity and mortality. Journal of Clinical Investigation, 2021, 131, .	3.9	72
53	Genetic mechanisms of critical illness in COVID-19. Nature, 2021, 591, 92-98.	13.7	1,014
54	Related expression of TRKA and P75 receptors and the changing copy number ofMYC-oncogenes determine the sensitivity of brain tumor cells to the treatment of the nerve growth factor in combination with cisplatin and temozolomide. Drug Metabolism and Personalized Therapy, 2021, .	0.3	1

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55	Autism Spectrum Disorders: Analysis of Mobile Elements at 7q11.23 Williamsâ€“Beuren Region by Comparative Genomics. Genes, 2021, 12, 1605.	1.0	1
56	Identification of a Novel Pathogenic Variant in the NAGLU Gene in a Child with Neurodevelopmental Delay. Journal of Autism and Developmental Disorders, 2021, , 1.	1.7	0
57	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. Frontiers in Oncology, 2021, 11, 649435.	1.3	2
58	Post-Mendelian Genetic Model in COVID-19. Cardiology and Cardiovascular Medicine, 2021, 05, .	0.1	10
59	JNK signaling provides a novel therapeutic target for Rett syndrome. BMC Biology, 2021, 19, 256.	1.7	6
60	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. International Journal of Molecular Sciences, 2021, 22, 13439.	1.8	23
61	Governance of Access in Biobanking: The Case of Telethon Network of Genetic Biobanks. Biopreservation and Biobanking, 2021, 19, 483-492.	0.5	1
62	Spondyloocular Syndrome: A Novel XYLT2 Variant with Description of the Neonatal Phenotype. Frontiers in Genetics, 2021, 12, 761264.	1.1	4
63	MEIS2 gene is responsible for intellectual disability, cardiac defects and a distinct facial phenotype. European Journal of Medical Genetics, 2020, 63, 103627.	0.7	23
64	Testing single/combined clinical categories on 5110 Italian patients with developmental phenotypes to improve arrayâ€based detection rate. Molecular Genetics & Genomic Medicine, 2020, 8, e1056.	0.6	6
65	New frontiers to cure Alport syndrome: COL4A3 and COL4A5 gene editing in podocyte-lineage cells. European Journal of Human Genetics, 2020, 28, 480-490.	1.4	22
66	Detection of Cryptic Mosaicism in X-linked Alport Syndrome Prompts to Re-evaluate Living-donor Kidney Transplantation. Transplantation, 2020, 104, 2360-2364.	0.5	4
67	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. Brain, 2020, 143, 2380-2387.	3.7	34
68	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	1.4	208
69	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. Brain, 2020, 143, 3564-3573.	3.7	23
70	RB1 Germline Variant Predisposing to a Rare Ovarian Germ Cell Tumor: A Case Report. Frontiers in Oncology, 2020, 10, 1467.	1.3	1
71	Vestibular and audiological findings in the Alport syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2345-2358.	0.7	6
72	Clinical Features at Onset and Genetic Characterization of Pediatric and Adult Patients with TNF-Î± Receptorâ€”Associated Periodic Syndrome (TRAPS): A Series of 80 Cases from the AIDA Network. Mediators of Inflammation, 2020, 2020, 1-12.	1.4	24

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73	A novel mutation in LMX1B gene in a newborn with nailâ€patella syndrome: Clinical and dermoscopic findings. Pediatric Dermatology, 2020, 37, 1205-1206.	0.5	2
74	X-Linked Alport Syndrome in Women: Genotype and Clinical Course in 24 Cases. Frontiers in Medicine, 2020, 7, 580376.	1.2	14
75	Cell-free DNA next-generation sequencing liquid biopsy as a new revolutionary approach for arteriovenous malformation. JVS Vascular Science, 2020, 1, 176-180.	0.4	17
76	High rate of HDR in gene editing of p.(Thr158Met) MECP2 mutational hotspot. European Journal of Human Genetics, 2020, 28, 1231-1242.	1.4	10
77	Assessment of haptoglobin alleles in autism spectrum disorders. Scientific Reports, 2020, 10, 7758.	1.6	2
78	AAV-mediated FOXG1 gene editing in human Rett primary cells. European Journal of Human Genetics, 2020, 28, 1446-1458.	1.4	12
79	Role of Colchicine Treatment in Tumor Necrosis Factor Receptor Associated Periodic Syndrome (TRAPS): Real-Life Data from the AIDA Network. Mediators of Inflammation, 2020, 2020, 1-6.	1.4	7
80	Gene replacement ameliorates deficits in mouse and human models of cyclin-dependent kinase-like 5 disorder. Brain, 2020, 143, 811-832.	3.7	34
81	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	1.2	14
82	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
83	Twoâ€pointâ€NGS analysis of cancer genes in cellâ€free DNA of metastatic cancer patients. Cancer Medicine, 2020, 9, 2052-2061.	1.3	8
84	An Italian family carrying a new mutation in the COL4A1 gene. Journal of the Neurological Sciences, 2020, 414, 116815.	0.3	0
85	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. Orphanet Journal of Rare Diseases, 2020, 15, 103.	1.2	23
86	17p13.3 microdeletion including YWHAE and CRK genes: towards a clinical characterization. Neurological Sciences, 2020, 41, 2259-2262.	0.9	9
87	Human CRY1 variants associate with attention deficit/hyperactivity disorder. Journal of Clinical Investigation, 2020, 130, 3885-3900.	3.9	35
88	Clinical and molecular characterization of COVID-19 hospitalized patients. PLoS ONE, 2020, 15, e0242534.	1.1	25
89	Related expression of TRKA and P75 receptors and the changing copy number ofMYC-oncogenes determine the sensitivity of brain tumor cells to the treatment of the nerve growth factor in combination with cisplatin and temozolomide. Drug Metabolism and Drug Interactions, 2020, 35, .	0.3	0
90	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189.	0.9	97

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91	A glomerulus-on-a-chip to recapitulate the human glomerular filtration barrier. Nature Communications, 2019, 10, 3656.	5.8	137
92	PIK3CA-CDKN2A clonal evolution in metastatic breast cancer and multiple points cell-free DNA analysis. Cancer Cell International, 2019, 19, 274.	1.8	1
93	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	1.0	63
94	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
95	Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of BRCA1/2 Variants of Uncertain Significance. Cancers, 2019, 11, 295.	1.7	24
96	Non-collagen genes role in digenic Alport syndrome. BMC Nephrology, 2019, 20, 70.	0.8	16
97	Agingâ€associated genes and letâ€7 microRNAs: a contribution to myogenic program dysregulation in oculopharyngeal muscular dystrophy. FASEB Journal, 2019, 33, 7155-7167.	0.2	19
98	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	0.8	23
99	Hints for Genetic and Clinical Differentiation of Adult-Onset Monogenic Autoinflammatory Diseases. Mediators of Inflammation, 2019, 2019, 1-29.	1.4	17
100	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127
101	Evidence of predisposing epimutation in retinoblastoma. Human Mutation, 2019, 40, 201-206.	1.1	16
102	Lowâ€level <scp>TP</scp>53 mutational load antecedes clonal expansion in chronic lymphocytic leukaemia. British Journal of Haematology, 2019, 184, 657-659.	1.2	2
103	Specific clonal expansion at disease progression (PD) in solid cancers pinpointed by cell free DNA analysis.. Journal of Clinical Oncology, 2019, 37, e13144-e13144.	0.8	0
104	Altered expression of RXFP1 receptor contributes to the inefficacy of relaxin-based anti-fibrotic treatments in systemic sclerosis. Clinical and Experimental Rheumatology, 2019, 37 Suppl 119, 69-75.	0.4	4
105	CKAP2L mutation confirms the diagnosis of Filippi syndrome. Clinical Genetics, 2018, 93, 1109-1110.	1.0	9
106	Parent-of-origin effect of hypomorphic pathogenic variants and somatic mosaicism impact on phenotypic expression of retinoblastoma. European Journal of Human Genetics, 2018, 26, 1026-1037.	1.4	19
107	Regulatory variants of FOXG1 in the context of its topological domain organisation. European Journal of Human Genetics, 2018, 26, 186-196.	1.4	20
108	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	1.1	67

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109	Alport syndrome: a unified classification of genetic disorders of collagen IV Î±345: a position paper of the Alport Syndrome Classification Working Group. Kidney International, 2018, 93, 1045-1051.	2.6	206
110	Germline mutations in lung cancer and personalized medicine. Familial Cancer, 2018, 17, 429-430.	0.9	5
111	Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy. Clinical Dysmorphology, 2018, 27, 18-20.	0.1	7
112	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. Human Mutation, 2018, 39, 302-314.	1.1	16
113	Commentary: Potential Links between Hepadnavirus and Bornavirus Sequences in the Host Genome and Cancer. Frontiers in Microbiology, 2018, 9, 1649.	1.5	0
114	Functional Connectivity and Genetic Profile of a â€œDouble-Cortexâ€•Like Malformation. Frontiers in Integrative Neuroscience, 2018, 12, 22.	1.0	11
115	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated Î±-tubulin defect which improves after iHDAC6 treatment in Rett syndrome. Experimental Cell Research, 2018, 368, 225-235.	1.2	36
116	Omic Approach in Non-smoker Female with Lung Squamous Cell Carcinoma Pinpoints to Germline Susceptibility and Personalized Medicine. Cancer Research and Treatment, 2018, 50, 356-365.	1.3	20
117	AB0185â€…Altered expression of relaxin receptor rxfp1/lgr7 in dermal fibroblasts contributes to the inefficacy of relaxin-based anti-fibrotic treatments in systemic sclerosis. , 2018, , .		0
118	Advances and unmet needs in genetic, basic and clinical science in Alport syndrome: report from the 2015 International Workshop on Alport Syndrome. Nephrology Dialysis Transplantation, 2017, 32, gfw095.	0.4	40
119	Inherited human IRAK-1 deficiency selectively impairs TLR signaling in fibroblasts. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E514-E523.	3.3	49
120	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	1.1	13
121	Combined ultrasound and exome sequencing approach recognizes Opitz G/BBB syndrome in two malformed fetuses. Clinical Dysmorphology, 2017, 26, 18-25.	0.1	3
122	Alport syndrome: impact of digenic inheritance in patients management. Clinical Genetics, 2017, 92, 34-44.	1.0	52
123	De novo microdeletions and point mutations affecting SOX2 in three individuals with intellectual disability but without major eye malformations. American Journal of Medical Genetics, Part A, 2017, 173, 435-443.	0.7	19
124	Intersociety policy statement on the use of whole-exome sequencing in the critically ill newborn infant. Italian Journal of Pediatrics, 2017, 43, 100.	1.0	51
125	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. Genome Medicine, 2017, 9, 67.	3.6	29
126	Potentially Treatable Disorder Diagnosed Post Mortem by Exome Analysis in a Boy with Respiratory Distress. International Journal of Molecular Sciences, 2016, 17, 306.	1.8	5

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127	A Genome Wide Copy Number Variations Analysis in Autism Spectrum Disorder (Asd) and Intellectual Disability (Id) in Italian Families. Journal of Genetic Syndromes & Gene Therapy, 2016, 7, .	0.2	3
128	Nicolaidesâ€“Baraitser syndrome: defining a phenotype. Journal of Neurology, 2016, 263, 1659-1660.	1.8	4
129	Exome sequencing analysis in a pair of monozygotic twins re-evaluates the genetics behind their intellectual disability and reveals a CHD2 mutation. Brain and Development, 2016, 38, 590-596.	0.6	11
130	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. Molecular Genetics and Metabolism, 2016, 119, 214-222.	0.5	21
131	Exploiting the potential of next-generation sequencing in genomic medicine. Expert Review of Molecular Diagnostics, 2016, 16, 1037-1047.	1.5	5
132	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. European Journal of Medical Genetics, 2016, 59, 436-443.	0.7	20
133	The alliance between genetic biobanks and patient organisations: the experience of the telethon network of genetic biobanks. Orphanet Journal of Rare Diseases, 2016, 11, 142.	1.2	40
134	Clonality Analysis of Immunoglobulin Gene Rearrangement by Next-Generation Sequencing in Endemic Burkitt Lymphoma Suggests Antigen Drive Activation of BCR as Opposed to Sporadic Burkitt Lymphoma. American Journal of Clinical Pathology, 2016, 145, 116-127.	0.4	35
135	Visual impairment in FOXG1-mutated individuals and mice. Neuroscience, 2016, 324, 496-508.	1.1	41
136	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. Journal of Human Genetics, 2016, 61, 95-101.	1.1	29
137	Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1+/â^' patients and in foxg1+/â^' mice. European Journal of Human Genetics, 2016, 24, 871-880.	1.4	54
138	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. European Journal of Human Genetics, 2016, 24, 252-257.	1.4	10
139	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	0.7	22
140	Response to Phelan K. et al.: Letter to the Editor Regarding Disciglio et al: Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1681-1681.	0.7	2
141	Antiepileptic drugs in Rett Syndrome. European Journal of Paediatric Neurology, 2015, 19, 446-452.	0.7	13
142	Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204.	1.1	161
143	Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes are a common well recognizable cause of intellectual disability. Brain and Development, 2015, 37, 527-536.	0.6	32
144	Evidence of digenic inheritance in Alport syndrome. Journal of Medical Genetics, 2015, 52, 163-174.	1.5	129

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145	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. European Journal of Human Genetics, 2015, 23, 1523-1530.	1.4	37
146	Alteration of serum lipid profile, SRB1 loss, and impaired Nrf2 activation in CDKL5 disorder. Free Radical Biology and Medicine, 2015, 86, 156-165.	1.3	19
147	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. European Journal of Human Genetics, 2015, 23, 1116-1123.	1.4	63
148	FANCMc.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	1.4	91
149	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
150	Sporadic hereditary motor and sensory neuropathies: Advances in the diagnosis using next generation sequencing technology. Journal of the Neurological Sciences, 2015, 359, 409-417.	0.3	3
151	Epilepsy in Rett syndromeâ€"Lessons from the Rett networked database. Epilepsia, 2015, 56, 569-576.	2.6	47
152	Bone marrow failure and developmental delay caused by mutations in poly(A)-specific ribonuclease (PARN). Journal of Medical Genetics, 2015, 52, 738-748.	1.5	71
153	Dropped-head in recessive oculopharyngeal muscular dystrophy. Neuromuscular Disorders, 2015, 25, 869-872.	0.3	10
154	GluD1 is a common altered player in neuronal differentiation from both MECP2-mutated and CDKL5-mutated iPS cells. European Journal of Human Genetics, 2015, 23, 195-201.	1.4	65
155	Bone Marrow Failure and Developmental Delay Caused By Mutations in Poly(A)-Specific Ribonuclease. Blood, 2015, 126, 2404-2404.	0.6	11
156	Redox Imbalance and Morphological Changes in Skin Fibroblasts in Typical Rett Syndrome. Oxidative Medicine and Cellular Longevity, 2014, 2014, 1-10.	1.9	44
157	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
158	First Identification of a Triple Corneal Dystrophy Association: Keratoconus, Epithelial Basement Membrane Corneal Dystrophy and Fuchs' Endothelial Corneal Dystrophy. Case Reports in Ophthalmology, 2014, 5, 281-288.	0.3	24
159	Unbiased next generation sequencing analysis confirms the existence of autosomal dominant Alport syndrome in a relevant fraction of cases. Clinical Genetics, 2014, 86, 252-257.	1.0	121
160	9q31.1q31.3 deletion in two patients with similar clinical features: A newly recognized microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2014, 164, 685-690.	0.7	9
161	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. European Journal of Medical Genetics, 2014, 57, 163-168.	0.7	11
162	Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1666-1676.	0.7	49

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163	Capping of the N-terminus of PSD-95 by calmodulin triggers its postsynaptic release. EMBO Journal, 2014, 33, 1341-53.	3.5	64
164	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. Lung Cancer, 2014, 85, 168-174.	0.9	30
165	Superselective ophthalmic artery infusion of melphalan for intraocular retinoblastoma: preliminary results from 140 treatments. Acta Ophthalmologica, 2013, 91, 335-342.	0.6	54
166	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. Orphanet Journal of Rare Diseases, 2013, 8, 129.	1.2	39
167	Ambiguous external genitalia due to defect of 5-Î±-reductase in seven Iraqi patients: Prevalence of a novel mutation. Gene, 2013, 526, 490-493.	1.0	16
168	Exome sequencing overrides formal genetics: <scp>ASPM</scp> mutations in a case study of apparent Xâ€linked microcephalic intellectual deficit. Clinical Genetics, 2013, 83, 288-290.	1.0	9
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