Alessandra Renieri

List of Publications by Year in Descending Order

323 papers	11,923 citations	52 h-index	98 g-index
362 ext. papers	15,078 ext. citations	5.6 avg, IF	5.38 L-index

#	Paper	IF	Citations
323	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis.. *Neurological Sciences*, **2022**, 43, 2849	3.5	
322	Development and Implementation of the AIDA International Registry for Patients With Still's Disease.. *Frontiers in Medicine*, **2022**, 9, 878797	4.9	0
321	Identification of a Novel Pathogenic Variant in a Patient with a Neurodevelopmental Disorder.. *Genes*, **2022**, 13,	4.2	1
320	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. *Human Genetics*, **2021**, 141, 147	6.3	3
319	The polymorphism L412F in inhibits autophagy and is a marker of severe COVID-19 in males.. *Autophagy*, **2021**, 1-11	10.2	5
318	Exome sequencing in BRCA1-2 candidate familias: the contribution of other cancer susceptibility genes. *Frontiers in Oncology*, **2021**, 11, 649435	5.3	2
317	Identification of a Novel Pathogenic Variant in the NAGLU Gene in a Child with Neurodevelopmental Delay. *Journal of Autism and Developmental Disorders*, **2021**, 1	4.6	
316	Genetic mechanisms of critical illness in COVID-19. *Nature*, **2021**, 591, 92-98	50.4	451
315	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. *EBioMedicine*, **2021**, 65, 103246	8.8	25
314	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,	8.9	51
313	Age-dependent impact of the major common genetic risk factor for COVID-19 on severity and mortality **2021**,		5
312	Protective Role of a Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. *Genes*, **2021**, 12,	4.2	14
311	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	5.3	14
310	Solving unsolved rare neurological diseases-a Solve-RD viewpoint. *European Journal of Human Genetics*, **2021**, 29, 1332-1336	5.3	0
309	Drug survival of anakinra and canakinumab in monogenic autoinflammatory diseases: observational study from the International AIDA Registry. *Rheumatology*, **2021**, 60, 5705-5712	3.9	1
308	The phenomenon of multidrug resistance in glioblastomas. *Hematology/ Oncology and Stem Cell Therapy*, **2021**,	2.7	1
307	In response to the letter to the editor by Soha Ghanian et al. re our publication "Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males". *EBioMedicine*, **2021**, 68, 103426	8.8	

306	Common and rare variant analyses combined with single-cell multiomics reveal cell-type-specific molecular mechanisms of COVID-19 severity **2021**,		1
305	Severe COVID-19 in Hospitalized Carriers of Single Pathogenic Variants. *Journal of Personalized Medicine*, **2021**, 11,	3.6	5
304	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. *European Journal of Human Genetics*, **2021**, 29, 1325-1331	5.3	10
303	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	5.3	2
302	Intermediate Repeats Confer Genetic Risk for Severe COVID-19 Pneumonia Independently of Age. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	7
301	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. *European Journal of Human Genetics*, **2021**, 29, 1337-1347	5.3	4
300	Vitamin D and COVID-19 susceptibility and severity in the COVID-19 Host Genetics Initiative: A Mendelian randomization study. *PLoS Medicine*, **2021**, 18, e1003605	11.6	32
299	Clinical, molecular and glycophenotype insights in SLC39A8-CDG. *Orphanet Journal of Rare Diseases*, **2021**, 16, 307	4.2	0
298	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. *Brain Sciences*, **2021**, 11,	3.4	5
297	Biotechnological Agents for Patients With Tumor Necrosis Factor Receptor Associated Periodic Syndrome-Therapeutic Outcome and Predictors of Response: Real-Life Data From the AIDA Network. *Frontiers in Medicine*, **2021**, 8, 668173	4.9	3
296	A pilot study of next generation sequencing-liquid biopsy on cell-free DNA as a novel non-invasive diagnostic tool for Klippel-Trenaunay syndrome. *Vascular*, **2021**, 29, 85-91	1.3	6
295	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	7.8	4
294	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	3.2	2
293	CDKL5 mutations may mimic Pitt-Hopkins syndrome phenotype. *European Journal of Medical Genetics*, **2021**, 64, 104102	2.6	
292	A new mutation in DNM2 gene in a large Italian family. *Neurological Sciences*, **2021**, 42, 2509-2513	3.5	
291	IQSEC2 disorder: A new disease entity or a Rett spectrum continuum?. *Clinical Genetics*, **2021**, 99, 462-474	4	3
290	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	5.3	20
289	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, 1759720X211037178	3.8	0

288	Epilepsy in Nicolaides-Baraitser Syndrome: Review of Literature and Report of 25 Patients Focusing on Treatment Aspects. *Neuropediatrics*, **2021**, 52, 109-122	1.6	1
287	Mapping the human genetic architecture of COVID-19. *Nature*, **2021**,	50.4	162
286	SELP Asp603Asn and severe thrombosis in COVID-19 males. *Journal of Hematology and Oncology*, **2021**, 14, 123	22.4	3
285	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. *Journal of Neurology*, **2021**, 1	5.5	3
284	Novel retinal finding in a patient with 4q12 deletion. *Ophthalmic Genetics*, **2021**, 1-3	1.2	
283	Age-dependent impact of the major common genetic risk factor for COVID-19 on severity and mortality. *Journal of Clinical Investigation*, **2021**, 131,	15.9	15
282	JNK signaling provides a novel therapeutic target for Rett syndrome.. *BMC Biology*, **2021**, 19, 256	7.3	3
281	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients.. *Genes and Immunity*, **2021**,	4.4	4
280	New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing.. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
279	Spondyloocular Syndrome: A Novel Variant with Description of the Neonatal Phenotype.. *Frontiers in Genetics*, **2021**, 12, 761264	4.5	1
278	Guidelines for Genetic Testing and Management of Alport Syndrome.. *Clinical Journal of the American Society of Nephrology: CJASN*, **2021**,	6.9	5
277	Cell-free DNA next-generation sequencing liquid biopsy as a new revolutionary approach for arteriovenous malformation. *JVS Vascular Science*, **2020**, 1, 176-180	1.3	4
276	High rate of HDR in gene editing of p.(Thr158Met) MECP2 mutational hotspot. *European Journal of Human Genetics*, **2020**, 28, 1231-1242	5.3	2
275	Assessment of haptoglobin alleles in autism spectrum disorders. *Scientific Reports*, **2020**, 10, 7758	4.9	1
274	AAV-mediated FOXG1 gene editing in human Rett primary cells. *European Journal of Human Genetics*, **2020**, 28, 1446-1458	5.3	6
273	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, 1936960	4.3	5
272	Gene replacement ameliorates deficits in mouse and human models of cyclin-dependent kinase-like 5 disorder. *Brain*, **2020**, 143, 811-832	11.2	11
271	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. *Journal of Molecular Diagnostics*, **2020**, 22, 1205-1215	5.1	8

270	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell*, **2020**, 180, 568-584.e23	56.2	578
269	Two-point-NGS analysis of cancer genes in cell-free DNA of metastatic cancer patients. *Cancer Medicine*, **2020**, 9, 2052-2061	4.8	7
268	An Italian family carrying a new mutation in the COL4A1 gene. *Journal of the Neurological Sciences*, **2020**, 414, 116815	3.2	
267	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	4.2	14
266	Human CRY1 variants associate with attention deficit/hyperactivity disorder. *Journal of Clinical Investigation*, **2020**, 130, 3885-3900	15.9	15
265	Clinical and molecular characterization of COVID-19 hospitalized patients. *PLoS ONE*, **2020**, 15, e0242534	3.7	14
264	Related expression of TRKA and P75 receptors and the changing copy number of MYC-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*, **2020**,	2	1
263	Testing single/combined clinical categories on 5110 Italian patients with developmental phenotypes to improve array-based detection rate. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1056	2.3	6
262	New frontiers to cure Alport syndrome: COL4A3 and COL4A5 gene editing in podocyte-lineage cells. *European Journal of Human Genetics*, **2020**, 28, 480-490	5.3	10
261	Detection of Cryptic Mosaicism in X-linked Alport Syndrome Prompts to Re-evaluate Living-donor Kidney Transplantation. *Transplantation*, **2020**, 104, 2360-2364	1.8	4
260	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. *Brain*, **2020**, 143, 2380-2387	11.2	15
259	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	5.3	132
258	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. *Brain*, **2020**, 143, 3564-3573	11.2	7
257	Germline Variant Predisposing to a Rare Ovarian Germ Cell Tumor: A Case Report. *Frontiers in Oncology*, **2020**, 10, 1467	5.3	1
256	Vestibular and audiological findings in the Alport syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2345-2358	2.5	0
255	Clinical Features at Onset and Genetic Characterization of Pediatric and Adult Patients with TNF-Receptor-Associated Periodic Syndrome (TRAPS): A Series of 80 Cases from the AIDA Network. *Mediators of Inflammation*, **2020**, 2020, 8562485	4.3	13
254	A novel mutation in LMX1B gene in a newborn with nail-patella syndrome: Clinical and dermoscopic findings. *Pediatric Dermatology*, **2020**, 37, 1205-1206	1.9	1
253	X-Linked Alport Syndrome in Women: Genotype and Clinical Course in 24 Cases. *Frontiers in Medicine*, **2020**, 7, 580376	4.9	2

252	MEIS2 gene is responsible for intellectual disability, cardiac defects and a distinct facial phenotype. *European Journal of Medical Genetics*, **2020**, 63, 103627	2.6	10
251	17p13.3 microdeletion including YWHAE and CRK genes: towards a clinical characterization. *Neurological Sciences*, **2020**, 41, 2259-2262	3.5	4
250	Diagnosis and management in Pitt-Hopkins syndrome: First international consensus statement. *Clinical Genetics*, **2019**, 95, 462-478	4	34
249	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
248	Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of Variants of Uncertain Significance. *Cancers*, **2019**, 11,	6.6	16
247	Non-collagen genes role in digenic Alport syndrome. *BMC Nephrology*, **2019**, 20, 70	2.7	9
246	Aging-associated genes and microRNAs: a contribution to myogenic program dysregulation in oculopharyngeal muscular dystrophy. *FASEB Journal*, **2019**, 33, 7155-7167	0.9	6
245	Analysis of the Phenotypes in the Rett Networked Database. *International Journal of Genomics*, **2019**, 2019, 6956934	2.5	14
244	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. *Pediatric Nephrology*, **2019**, 34, 1175-1189	3.2	70
243	A glomerulus-on-a-chip to recapitulate the human glomerular filtration barrier. *Nature Communications*, **2019**, 10, 3656	17.4	66
242	PIK3CA-CDKN2A clonal evolution in metastatic breast cancer and multiple points cell-free DNA analysis. *Cancer Cell International*, **2019**, 19, 274	6.4	0
241	Specific clonal expansion at disease progression (PD) in solid cancers pinpointed by cell free DNA analysis.. *Journal of Clinical Oncology*, **2019**, 37, e13144-e13144	2.2	
240	Hints for Genetic and Clinical Differentiation of Adult-Onset Monogenic Autoinflammatory Diseases. *Mediators of Inflammation*, **2019**, 2019, 3293145	4.3	7
239	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. *Genetics in Medicine*, **2019**, 21, 816-825	8.1	71
238	Evidence of predisposing epimutation in retinoblastoma. *Human Mutation*, **2019**, 40, 201-206	4.7	14
237	Low-level TP53 mutational load antecedes clonal expansion in chronic lymphocytic leukaemia. *British Journal of Haematology*, **2019**, 184, 657-659	4.5	2
236	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	2.2	4
235	CKAP2L mutation confirms the diagnosis of Filippi syndrome. *Clinical Genetics*, **2018**, 93, 1109-1110	4	6

234	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	5.3	11
233	Regulatory variants of FOXG1 in the context of its topological domain organisation. *European Journal of Human Genetics*, **2018**, 26, 186-196	5.3	9
232	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. *Genetics in Medicine*, **2018**, 20, 965-975	8.1	37
231	Alport syndrome: a unified classification of genetic disorders of collagen IV α345: a position paper of the Alport Syndrome Classification Working Group. *Kidney International*, **2018**, 93, 1045-1051	9.9	108
230	Germline mutations in lung cancer and personalized medicine. *Familial Cancer*, **2018**, 17, 429-430	3	4
229	Functional Connectivity and Genetic Profile of a "Double-Cortex"-Like Malformation. *Frontiers in Integrative Neuroscience*, **2018**, 12, 22	3.2	5
228	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated α-tubulin defect which improves after iHDAC6 treatment in Rett syndrome. *Experimental Cell Research*, **2018**, 368, 225-235	4.2	31
227	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	5.2	12
226	Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy. *Clinical Dysmorphology*, **2018**, 27, 18-20	0.9	4
225	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. *Human Mutation*, **2018**, 39, 302-314	4.7	11
224	Commentary: Potential Links between Hepadnavirus and Bornavirus Sequences in the Host Genome and Cancer. *Frontiers in Microbiology*, **2018**, 9, 1649	5.7	
223	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	11.5	31
222	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. *Genetics in Medicine*, **2017**, 19, 701-710	8.1	4
221	Intersociety policy statement on the use of whole-exome sequencing in the critically ill newborn infant. *Italian Journal of Pediatrics*, **2017**, 43, 100	3.2	41
220	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. *Genome Medicine*, **2017**, 9, 67	14.4	17
219	Combined ultrasound and exome sequencing approach recognizes Opitz G/BBB syndrome in two malformed fetuses. *Clinical Dysmorphology*, **2017**, 26, 18-25	0.9	3
218	Alport syndrome: impact of digenic inheritance in patients management. *Clinical Genetics*, **2017**, 92, 34-44	4	33
217	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	2.5	14

216	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, 916-924	4.3	31
215	Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1(+/-) patients and in foxg1(+/-) mice. *European Journal of Human Genetics*, **2016**, 24, 871-80	5.3	39
214	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. *European Journal of Human Genetics*, **2016**, 24, 252-7	5.3	9
213	Exploiting the potential of next-generation sequencing in genomic medicine. *Expert Review of Molecular Diagnostics*, **2016**, 16, 1037-47	3.8	4
212	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. *European Journal of Medical Genetics*, **2016**, 59, 436-43	2.6	16
211	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	4.2	14
210	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, 116-27	1.9	26
209	Visual impairment in FOXG1-mutated individuals and mice. *Neuroscience*, **2016**, 324, 496-508	3.9	27
208	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. *Journal of Human Genetics*, **2016**, 61, 95-101	4.3	18
207	Potentially Treatable Disorder Diagnosed Post Mortem by Exome Analysis in a Boy with Respiratory Distress. *International Journal of Molecular Sciences*, **2016**, 17, 306	6.3	5
206	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,		2
205	Nicolaides-Baraitser syndrome: defining a phenotype. *Journal of Neurology*, **2016**, 263, 1659-60	5.5	4
204	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, 590-6	2.2	10
203	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. *Molecular Genetics and Metabolism*, **2016**, 119, 214-222	3.7	19
202	Evidence of digenic inheritance in Alport syndrome. *Journal of Medical Genetics*, **2015**, 52, 163-74	5.8	95
201	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. *European Journal of Human Genetics*, **2015**, 23, 1523-30	5.3	33
200	Alteration of serum lipid profile, SRB1 loss, and impaired Nrf2 activation in CDKL5 disorder. *Free Radical Biology and Medicine*, **2015**, 86, 156-65	7.8	15
199	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. *European Journal of Human Genetics*, **2015**, 23, 1116-23	5.3	49

198	FANCM c.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, 5345-55	5.6	68
197	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. *American Journal of Human Genetics*, **2015**, 96, 784-96	11	35
196	Sporadic hereditary motor and sensory neuropathies: Advances in the diagnosis using next generation sequencing technology. *Journal of the Neurological Sciences*, **2015**, 359, 409-17	3.2	3
195	Epilepsy in Rett syndrome--lessons from the Rett networked database. *Epilepsia*, **2015**, 56, 569-76	6.4	30
194	Bone marrow failure and developmental delay caused by mutations in poly(A)-specific ribonuclease (PARN). *Journal of Medical Genetics*, **2015**, 52, 738-48	5.8	56
193	Dropped-head in recessive oculopharyngeal muscular dystrophy. *Neuromuscular Disorders*, **2015**, 25, 869-72	2.9	8
192	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	5.3	56
191	Recurrent duplications of 17q12 associated with variable phenotypes. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3038-45	2.5	17
190	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	2.5	1
189	Antiepileptic drugs in Rett Syndrome. *European Journal of Paediatric Neurology*, **2015**, 19, 446-52	3.8	7
188	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204	4.7	122
187	Coffin-Siris and Nicolaides-Baraitser syndromes are a common well recognizable cause of intellectual disability. *Brain and Development*, **2015**, 37, 527-36	2.2	26
186	Bone Marrow Failure and Developmental Delay Caused By Mutations in Poly(A)-Specific Ribonuclease. *Blood*, **2015**, 126, 2404-2404	2.2	
185	9q31.1q31.3 deletion in two patients with similar clinical features: a newly recognized microdeletion syndrome?. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 685-90	2.5	6
184	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. *European Journal of Medical Genetics*, **2014**, 57, 163-8	2.6	10
183	Interstitial 22q13 deletions not involving SHANK3 gene: a new contiguous gene syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1666-76	2.5	36
182	Capping of the N-terminus of PSD-95 by calmodulin triggers its postsynaptic release. *EMBO Journal*, **2014**, 33, 1341-53	13	49
181	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. *Lung Cancer*, **2014**, 85, 168-74	5.9	21

180	Redox imbalance and morphological changes in skin fibroblasts in typical Rett syndrome. *Oxidative Medicine and Cellular Longevity*, **2014**, 2014, 195935	6.7	36
179	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
178	First identification of a triple corneal dystrophy association: keratoconus, epithelial basement membrane corneal dystrophy and fuchsŒendothelial corneal dystrophy. *Case Reports in Ophthalmology*, **2014**, 5, 281-8	0.7	17
177	Unbiased next generation sequencing analysis confirms the existence of autosomal dominant Alport syndrome in a relevant fraction of cases. *Clinical Genetics*, **2014**, 86, 252-7	4	92
176	Superselective ophthalmic artery infusion of melphalan for intraocular retinoblastoma: preliminary results from 140 treatments. *Acta Ophthalmologica*, **2013**, 91, 335-42	3.7	46
175	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. *Orphanet Journal of Rare Diseases*, **2013**, 8, 129	4.2	28
174	Ambiguous external genitalia due to defect of 5-□reductase in seven Iraqi patients: prevalence of a novel mutation. *Gene*, **2013**, 526, 490-3	3.8	11
173	Exome sequencing overrides formal genetics: ASPM mutations in a case study of apparent X-linked microcephalic intellectual deficit. *Clinical Genetics*, **2013**, 83, 288-90	4	8
172	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. *Orphanet Journal of Rare Diseases*, **2013**, 8, 63	4.2	47
171	A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. *Human Molecular Genetics*, **2013**, 22, 5121-35	5.6	138
170	Variant of Rett Syndrome and CDKL5 Gene: Clinical and Autonomic Description of 10 Cases. *Neuropediatrics*, **2013**, 44, 237-238	1.6	1
169	Prognostic value of glomerular collagen IV immunofluorescence studies in male patients with X-linked Alport syndrome. *Clinical Journal of the American Society of Nephrology: CJASN*, **2013**, 8, 749-55	6.9	15
168	The role of surgical lung biopsy in the management of interstitial lung disease: experience from a single institution in the UK. *Interactive Cardiovascular and Thoracic Surgery*, **2013**, 17, 253-7	1.8	32
167	Dual copy number variants involving 16p11 and 6q22 in a case of childhood apraxia of speech and pervasive developmental disorder. *European Journal of Human Genetics*, **2013**, 21, 361-5	5.3	31
166	Revealing the complexity of a monogenic disease: rett syndrome exome sequencing. *PLoS ONE*, **2013**, 8, e56599	3.7	45
165	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. *Clinical Genetics*, **2012**, 82, 248-55	4	104
164	Expanding the phenotype associated with FOXG1 mutations and in vivo FoxG1 chromatin-binding dynamics. *Clinical Genetics*, **2012**, 82, 395-403	4	18
163	Xq28 duplications including MECP2 in five females: Expanding the phenotype to□severe mental retardation. *European Journal of Medical Genetics*, **2012**, 55, 404-13	2.6	38

162	A unique patient presenting with concomitant Klinefelter syndrome, Alport syndrome, and craniopharyngioma. *Journal of Andrology*, **2012**, 33, 1155-9		7
161	13q deletion syndrome and retinoblastoma in identical dichorionic diamniotic monozygotic twins. *European Journal of Ophthalmology*, **2012**, 22, 857-60	1.9	3
160	Rett networked database: an integrated clinical and genetic network of Rett syndrome databases. *Human Mutation*, **2012**, 33, 1031-6	4.7	14
159	Periventricular heterotopia with white matter abnormalities associated with 6p25 deletion. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1793-7	2.5	20
158	Epigenetic and copy number variation analysis in retinoblastoma by MS-MLPA. *Pathology and Oncology Research*, **2012**, 18, 703-12	2.6	37
157	Variant of Rett syndrome and CDKL5 gene: clinical and autonomic description of 10 cases. *Neuropediatrics*, **2012**, 43, 37-43	1.6	22
156	Reduced expression of MECP2 affects cell commitment and maintenance in neurons by triggering senescence: new perspective for Rett syndrome. *Molecular Biology of the Cell*, **2012**, 23, 1435-45	3.5	31
155	Advances in Alport syndrome diagnosis using next-generation sequencing. *European Journal of Human Genetics*, **2012**, 20, 50-7	5.3	66
154	Phosphatase and tensin homolog (PTEN) gene mutations and autism: literature review and a case report of a patient with Cowden syndrome, autistic disorder, and epilepsy. *Journal of Child Neurology*, **2012**, 27, 392-7	2.5	52
153	Vav1 haploinsufficiency in a common variable immunodeficiency patient with defective T-cell function. *International Journal of Immunopathology and Pharmacology*, **2012**, 25, 811-7	3	15
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5	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in males		1
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