Alessandra Renieri

List of Publications by Citations

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	Rett syndrome: revised diagnostic criteria and nomenclature. *Annals of Neurology*, **2010**, 68, 944-50	9.4	804
322	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
321	Genetic mechanisms of critical illness in COVID-19. *Nature*, **2021**, 591, 92-98	50.4	451
320	X-linked Alport syndrome: natural history in 195 families and genotype- phenotype correlations in males. *Journal of the American Society of Nephrology: JASN*, **2000**, 11, 649-657	12.7	332
319	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
318	FOXG1 is responsible for the congenital variant of Rett syndrome. *American Journal of Human Genetics*, **2008**, 83, 89-93	11	312
317	X-linked Alport syndrome: natural history and genotype-phenotype correlations in girls and women belonging to 195 families: a "European Community Alport Syndrome Concerted Action" study. *Journal of the American Society of Nephrology: JASN*, **2003**, 14, 2603-10	12.7	298
316	CDKL5 belongs to the same molecular pathway of MeCP2 and it is responsible for the early-onset seizure variant of Rett syndrome. *Human Molecular Genetics*, **2005**, 14, 1935-46	5.6	248
315	MYH9-related disease: May-Hegglin anomaly, Sebastian syndrome, Fechtner syndrome, and Epstein syndrome are not distinct entities but represent a variable expression of a single illness. *Medicine (United States)*, **2003**, 82, 203-15	1.8	203
314	A mutation in the rett syndrome gene, MECP2, causes X-linked mental retardation and progressive spasticity in males. *American Journal of Human Genetics*, **2000**, 67, 982-5	11	196
313	CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms. *Journal of Medical Genetics*, **2005**, 42, 103-7	5.8	180
312	Cloning of the gene for ocular albinism type 1 from the distal short arm of the X chromosome. *Nature Genetics*, **1995**, 10, 13-9	36.3	171
311	Mapping the human genetic architecture of COVID-19. *Nature*, **2021**,	50.4	162
310	COL4A3/COL4A4 mutations: from familial hematuria to autosomal-dominant or recessive Alport syndrome. *Kidney International*, **2002**, 61, 1947-56	9.9	143
309	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
308	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
307	FACL4, encoding fatty acid-CoA ligase 4, is mutated in nonspecific X-linked mental retardation. *Nature Genetics*, **2002**, 30, 436-40	36.3	123

306	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204	4.7	122
305	Revised nomenclature for the mammalian long-chain acyl-CoA synthetase gene family. *Journal of Lipid Research*, **2004**, 45, 1958-61	6.3	119
304	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
303	FACL4, a new gene encoding long-chain acyl-CoA synthetase 4, is deleted in a family with Alport syndrome, elliptocytosis, and mental retardation. *Genomics*, **1998**, 47, 350-8	4.3	106
302	Preserved speech variant is allelic of classic Rett syndrome. *European Journal of Human Genetics*, **2000**, 8, 325-30	5.3	105
301	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. *Clinical Genetics*, **2012**, 82, 248-55	4	104
300	Preserved speech variants of the Rett syndrome: molecular and clinical analysis. *American Journal of Medical Genetics Part A*, **2001**, 104, 14-22		100
299	Mutations in FN1 cause glomerulopathy with fibronectin deposits. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 2538-43	11.5	96
298	Evidence of digenic inheritance in Alport syndrome. *Journal of Medical Genetics*, **2015**, 52, 163-74	5.8	95
297	LINE-1 elements at the sites of molecular rearrangements in Alport syndrome-diffuse leiomyomatosis. *American Journal of Human Genetics*, **1999**, 64, 62-9	11	95
296	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
295	Real-time quantitative PCR as a routine method for screening large rearrangements in Rett syndrome: Report of one case of MECP2 deletion and one case of MECP2 duplication. *Human Mutation*, **2004**, 24, 172-7	4.7	92
294	Novel FOXG1 mutations associated with the congenital variant of Rett syndrome. *Journal of Medical Genetics*, **2010**, 47, 49-53	5.8	91
293	Autosomal-dominant Alport syndrome: natural history of a disease due to COL4A3 or COL4A4 gene. *Kidney International*, **2004**, 65, 1598-603	9.9	90
292	Deciphering the underlying genetic and epigenetic events leading to gastric carcinogenesis. *Journal of Cellular Physiology*, **2007**, 211, 287-95	7	89
291	Mowat-Wilson syndrome: facial phenotype changing with age: study of 19 Italian patients and review of the literature. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 417-26	2.5	83
290	A clinical, genetic, and biochemical characterization of SPG7 mutations in a large cohort of patients with hereditary spastic paraplegia. *Human Mutation*, **2008**, 29, 522-31	4.7	74
289	Analysis of the OA1 gene reveals mutations in only one-third of patients with X-linked ocular albinism. *Human Molecular Genetics*, **1995**, 4, 2319-25	5.6	73

288	Autosomal dominant Alport syndrome: molecular analysis of the COL4A4 gene and clinical outcome. *Nephrology Dialysis Transplantation*, **2009**, 24, 1464-71	4.3	71
287	iPS cells to model CDKL5-related disorders. *European Journal of Human Genetics*, **2011**, 19, 1246-55	5.3	71
286	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
285	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. *Pediatric Nephrology*, **2019**, 34, 1175-1189	3.2	70
284	KCNE1-like gene is deleted in AMME contiguous gene syndrome: identification and characterization of the human and mouse homologs. *Genomics*, **1999**, 60, 251-7	4.3	69
283	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
282	Mutation spectrum of MLL2 in a cohort of Kabuki syndrome patients. *Orphanet Journal of Rare Diseases*, **2011**, 6, 38	4.2	68
281	Diagnostic criteria for the Zappella variant of Rett syndrome (the preserved speech variant). *Brain and Development*, **2009**, 31, 208-16	2.2	68
280	X-linked Alport syndrome: an SSCP-based mutation survey over all 51 exons of the COL4A5 gene. *American Journal of Human Genetics*, **1996**, 58, 1192-204	11	68
279	Rett syndrome: the complex nature of a monogenic disease. *Journal of Molecular Medicine*, **2003**, 81, 346-54	5.5	67
278	A glomerulus-on-a-chip to recapitulate the human glomerular filtration barrier. *Nature Communications*, **2019**, 10, 3656	17.4	66
277	Advances in Alport syndrome diagnosis using next-generation sequencing. *European Journal of Human Genetics*, **2012**, 20, 50-7	5.3	66
276	Study of MECP2 gene in Rett syndrome variants and autistic girls. *American Journal of Medical Genetics Part A*, **2003**, 119B, 102-7		61
275	Epilepsy in Rett syndrome: clinical and genetic features. *Epilepsy and Behavior*, **2010**, 19, 296-300	3.2	60
274	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
273	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
272	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
271	Early-onset seizure variant of Rett syndrome: definition of the clinical diagnostic criteria. *Brain and Development*, **2010**, 32, 17-24	2.2	51

270	Private inherited microdeletion/microduplications: implications in clinical practice. *European Journal of Medical Genetics*, **2008**, 51, 409-16	2.6	51
269	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
268	A 3 Mb deletion in 14q12 causes severe mental retardation, mild facial dysmorphisms and Rett-like features. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1994-8	2.5	50
267	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
266	Capping of the N-terminus of PSD-95 by calmodulin triggers its postsynaptic release. *EMBO Journal*, **2014**, 33, 1341-53	13	49
265	Epstein-Barr virus and gastric cancer: data and unanswered questions. *International Journal of Cancer*, **1993**, 53, 898-901	7.5	49
264	Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis: a new X linked contiguous gene deletion syndrome?. *Journal of Medical Genetics*, **1998**, 35, 273-8	5.8	49
263	Ultrastructural and immunohistochemical findings in Alport© syndrome: a study of 108 patients from 97 Italian families with particular emphasis on COL4A5 gene mutation correlations. *Journal of the American Society of Nephrology: JASN*, **1998**, 9, 1023-31	12.7	48
262	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
261	Mutation analysis of the MECP2 gene in British and Italian Rett syndrome females. *Journal of Molecular Medicine*, **2001**, 78, 648-55	5.5	47
260	Superselective ophthalmic artery infusion of melphalan for intraocular retinoblastoma: preliminary results from 140 treatments. *Acta Ophthalmologica*, **2013**, 91, 335-42	3.7	46
259	2q24-q31 deletion: report of a case and review of the literature. *European Journal of Medical Genetics*, **2007**, 50, 21-32	2.6	46
258	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
257	Revealing the complexity of a monogenic disease: rett syndrome exome sequencing. *PLoS ONE*, **2013**, 8, e56599	3.7	45
256	Epstein syndrome: another renal disorder with mutations in the nonmuscle myosin heavy chain 9 gene. *Human Genetics*, **2002**, 110, 182-6	6.3	43
255	Intracellular levels of the LIS1 protein correlate with clinical and neuroradiological findings in patients with classical lissencephaly. *Annals of Neurology*, **1999**, 45, 154-161	9.4	43
254	De novo mutation in the COL4A5 gene converting glycine 325 to glutamic acid in Alport syndrome. *Human Molecular Genetics*, **1992**, 1, 127-9	5.6	43
253	PAX6 mutation in a family with aniridia, congenital ptosis, and mental retardation. *Clinical Genetics*, **2001**, 60, 151-4	4	42

252	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
251	MECP2 deletions and genotype-phenotype correlation in Rett syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2775-84	2.5	41
250	Chromosome 2 deletion encompassing the MAP2 gene in a patient with autism and Rett-like features. *Clinical Genetics*, **2003**, 64, 497-501	4	41
249	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
248	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
247	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. *Nephrology Dialysis Transplantation*, **2006**, 21, 665-71	4.3	38
246	Mutations in the myocilin gene in families with primary open-angle glaucoma and juvenile open-angle glaucoma. *JAMA Ophthalmology*, **2003**, 121, 1034-8		38
245	Identification and characterization of a highly conserved protein absent in the Alport syndrome (A), mental retardation (M), midface hypoplasia (M), and elliptocytosis (E) contiguous gene deletion syndrome (AMME). *Genomics*, **1999**, 55, 335-40	4.3	38
244	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. *Genetics in Medicine*, **2018**, 20, 965-975	8.1	37
243	Epigenetic and copy number variation analysis in retinoblastoma by MS-MLPA. *Pathology and Oncology Research*, **2012**, 18, 703-12	2.6	37
242	The first Italian family with tibial muscular dystrophy caused by a novel titin mutation. *Journal of Neurology*, **2010**, 257, 575-9	5.5	37
241	A third MRX family (MRX68) is the result of mutation in the long chain fatty acid-CoA ligase 4 (FACL4) gene: proposal of a rapid enzymatic assay for screening mentally retarded patients. *Journal of Medical Genetics*, **2003**, 40, 11-7	5.8	37
240	Evidence for genetic heterogeneity in benign familial hematuria. *American Journal of Nephrology*, **1999**, 19, 464-7	4.6	37
239	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
238	Redox imbalance and morphological changes in skin fibroblasts in typical Rett syndrome. *Oxidative Medicine and Cellular Longevity*, **2014**, 2014, 195935	6.7	36
237	14q12 Microdeletion syndrome and congenital variant of Rett syndrome. *European Journal of Medical Genetics*, **2009**, 52, 148-52	2.6	36
236	Seizures and electroencephalographic findings in CDKL5 mutations: case report and review. *Brain and Development*, **2007**, 29, 239-42	2.2	36
235	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

234	Diagnosis and management in Pitt-Hopkins syndrome: First international consensus statement. *Clinical Genetics*, **2019**, 95, 462-478	4	34
233	Partial silencing of methyl cytosine protein binding 2 (MECP2) in mesenchymal stem cells induces senescence with an increase in damaged DNA. *FASEB Journal*, **2010**, 24, 1593-603	0.9	34
232	Genomic differences between retinoma and retinoblastoma. *Acta Oncologica*, **2008**, 47, 1483-92	3.2	34
231	Next generation sequencing in sporadic retinoblastoma patients reveals somatic mosaicism. *European Journal of Human Genetics*, **2015**, 23, 1523-30	5.3	33
230	Alport syndrome: impact of digenic inheritance in patients management. *Clinical Genetics*, **2017**, 92, 34-44	4	33
229	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
228	Clinical and molecular characterization of a patient with a 2q31.2-32.3 deletion identified by array-CGH. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 858-65	2.5	32
227	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
226	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
225	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
224	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
223	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
222	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
221	CAG repeat expansion in an italian family with spinocerebellar ataxia type 2 (SCA2): a clinical and genetic study. *European Neurology*, **1998**, 40, 164-8	2.1	31
220	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
219	Epilepsy in Rett syndrome--lessons from the Rett networked database. *Epilepsia*, **2015**, 56, 569-76	6.4	30
218	Array comparative genomic hybridization in retinoma and retinoblastoma tissues. *Cancer Science*, **2009**, 100, 465-71	6.9	30
217	Refinement of the 12q14 microdeletion syndrome: primordial dwarfism and developmental delay with or without osteopoikilosis. *European Journal of Human Genetics*, **2009**, 17, 1141-7	5.3	30

216	Non-syndromic X-linked mental retardation: from a molecular to a clinical point of view. *Journal of Cellular Physiology*, **2005**, 204, 8-20	7	30
215	Cloning of a human homologue of the Xenopus laevis APX gene from the ocular albinism type 1 critical region. *Human Molecular Genetics*, **1995**, 4, 373-82	5.6	30
214	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
213	High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. *European Journal of Human Genetics*, **2010**, 18, 1133-40	5.3	28
212	The XLMR gene ACSL4 plays a role in dendritic spine architecture. *Neuroscience*, **2009**, 159, 657-69	3.9	28
211	. *Medicine (United States)*, **2003**, 82, 203-215	1.8	28
210	Pseudoxanthoma elasticum: Point mutations in the ABCC6 gene and a large deletion including also ABCC1 and MYH11. *Human Mutation*, **2001**, 18, 85	4.7	28
209	Alport syndrome caused by a 5Ωdeletion within the COL4A5 gene. *Human Genetics*, **1992**, 89, 120-1	6.3	28
208	Visual impairment in FOXG1-mutated individuals and mice. *Neuroscience*, **2016**, 324, 496-508	3.9	27
207	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
206	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
205	Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism. *Clinical Genetics*, **2003**, 63, 510-5	4	26
204	Unmasking of a Recessive SCARF2 Mutation by a 22q11.12 de novo Deletion in a Patient with Van den Ende-Gupta Syndrome. *Molecular Syndromology*, **2010**, 1, 239-245	1.5	25
203	A case report: bone marrow mesenchymal stem cells from a Rett syndrome patient are prone to senescence and show a lower degree of apoptosis. *Journal of Cellular Biochemistry*, **2008**, 103, 1877-85	4.7	25
202	Germline mosaicism in Rett syndrome identified by prenatal diagnosis. *Clinical Genetics*, **2005**, 67, 258-604		25
201	ACE2gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population		25
200	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. *EBioMedicine*, **2021**, 65, 103246	8.8	25
199	Investigation of modifier genes within copy number variations in Rett syndrome. *Journal of Human Genetics*, **2011**, 56, 508-15	4.3	23

198	Syndromic mental retardation with thrombocytopenia due to 21q22.11q22.12 deletion: Report of three patients. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1711-7	2.5	23
197	Italian Rett database and biobank. *Human Mutation*, **2007**, 28, 329-35	4.7	23
196	Mutational screening of the RB1 gene in Italian patients with retinoblastoma reveals 11 novel mutations. *Journal of Human Genetics*, **2006**, 51, 209-216	4.3	23
195	Epidermal basement membrane alpha 5(IV) expression in females with Alport syndrome and severity of renal disease. *Kidney International*, **2003**, 64, 1787-91	9.9	23
194	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. *Pediatric Nephrology*, **2011**, 26, 717-24	3.2	22
193	Variant of Rett syndrome and CDKL5 gene: clinical and autonomic description of 10 cases. *Neuropediatrics*, **2012**, 43, 37-43	1.6	22
192	Cohen syndrome resulting from a novel large intragenic COH1 deletion segregating in an isolated Greek island population. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2221-6	2.5	22
191	Multiple endocrine neoplasia type 2 syndromes may be associated with renal malformations. *Journal of Internal Medicine*, **2001**, 250, 37-42	10.8	22
190	Alport syndrome and mental retardation: clinical and genetic dissection of the contiguous gene deletion syndrome in Xq22.3 (ATS-MR). *Journal of Medical Genetics*, **2002**, 39, 359-65	5.8	22
189	Deletion spanning the 5Œends of both the COL4A5 and COL4A6 genes in a patient with Alport© syndrome and leiomyomatosis. *Human Mutation*, **1994**, 4, 195-8	4.7	22
188	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. *Lung Cancer*, **2014**, 85, 168-74	5.9	21
187	Thin glomerular basement membrane disease: clinical significance of a morphological diagnosis--a collaborative study of the Italian Renal Immunopathology Group. *Nephrology Dialysis Transplantation*, **2005**, 20, 545-51	4.3	21
186	Mild brachydactyly type A1 maps to chromosome 2q35-q36 and is caused by a novel IHH mutation in a three generation family. *Journal of Medical Genetics*, **2003**, 40, 132-5	5.8	21
185	MECP2 gene mutation analysis in the British and Italian Rett Syndrome patients: hot spot map of the most recurrent mutations and bioinformatic analysis of a new MECP2 conserved region. *Brain and Development*, **2001**, 23 Suppl 1, S246-50	2.2	21
184	Variability of clinical phenotype in a large Alport family with Gly 1143 Ser change of collagen alpha 5(IV)-chain. *Nephron*, **1994**, 67, 444-9	3.3	21
183	Periventricular heterotopia with white matter abnormalities associated with 6p25 deletion. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1793-7	2.5	20
182	Five patients with novel overlapping interstitial deletions in 8q22.2q22.3. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1857-64	2.5	20
181	Major COL4A5 gene rearrangements in patients with juvenile type Alport syndrome. *American Journal of Medical Genetics Part A*, **1995**, 59, 380-5		20

180	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
179	Delineation of the phenotype associated with 7q36.1q36.2 deletion: long QT syndrome, renal hypoplasia and mental retardation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1195-9	2.5	19
178	Small frameshift deletions within the COL4A5 gene in juvenile-onset Alport syndrome. *Human Genetics*, **1993**, 92, 417-20	6.3	19
177	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. *Molecular Genetics and Metabolism*, **2016**, 119, 214-222	3.7	19
176	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
175	Expanding the phenotype associated with FOXG1 mutations and in vivo FoxG1 chromatin-binding dynamics. *Clinical Genetics*, **2012**, 82, 395-403	4	18
174	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. *Journal of Human Genetics*, **2007**, 52, 1011-1017	4.3	18
173	Inheritance of a 38-kb fragment in apparently sporadic facioscapulohumeral muscular dystrophy. *Muscle and Nerve*, **1999**, 22, 1437-41	3.4	18
172	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. *Genome Medicine*, **2017**, 9, 67	14.4	17
171	Recurrent duplications of 17q12 associated with variable phenotypes. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3038-45	2.5	17
170	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
169	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
168	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
167	A 9.3 Mb microdeletion of 3q27.3q29 associated with psychomotor and growth delay, tricuspid valve dysplasia and bifid thumb. *European Journal of Medical Genetics*, **2009**, 52, 131-3	2.6	16
166	Expanding the phenotype of 22q11 deletion syndrome: the MURCS association. *Clinical Dysmorphology*, **2008**, 17, 13-17	0.9	16
165	Single base pair deletions in exons 39 and 42 of the COL4A5 gene in Alport syndrome. *Human Molecular Genetics*, **1994**, 3, 201-2	5.6	16
164	A novel frameshift deletion in type IV collagen alpha 5 gene in a juvenile-type Alport syndrome patient: an adenine deletion (2940/2943 del A) in exon 34 of COL4A5. *Human Mutation*, **1994**, 3, 386-90	4.7	16
163	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

162	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
161	Creatine transporter defect diagnosed by proton NMR spectroscopy in males with intellectual disability. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2446-52	2.5	15
160	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
159	Retinoblastoma and mental retardation microdeletion syndrome: clinical characterization and molecular dissection using array CGH. *Journal of Human Genetics*, **2007**, 52, 535-542	4.3	15
158	A 2.6 Mb deletion of 6q24.3-25.1 in a patient with growth failure, cardiac septal defect, thin upperlip and asymmetric dysmorphic ears. *European Journal of Medical Genetics*, **2007**, 50, 315-21	2.6	15
157	Mosaicism in Alport syndrome with genetic counselling. *Journal of Medical Genetics*, **2000**, 37, 717-9	5.8	15
156	Human CRY1 variants associate with attention deficit/hyperactivity disorder. *Journal of Clinical Investigation*, **2020**, 130, 3885-3900	15.9	15
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