Andrea Ciolfi

List of Publications by Year in Descending Order

75 papers	2,346 citations	27 h-index	47 g-index
84 ext. papers	3,155 ext. citations	7.4 avg, IF	4.27 L-index

#	Paper	IF	Citations
75	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1
74	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. *American Journal of Human Genetics*, **2021**, 108, 2112-2129	11	2
73	Neonatal Manifestations of Chronic Granulomatous Disease: MAS/HLH and Necrotizing Pneumonia as Unusual Phenotypes and Review of the Literature. *Journal of Clinical Immunology*, **2021**, 1	5.7	1
72	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	11	12
71	Co-occurring SYNJ1 and SHANK3 variants in a girl with intellectual disability, early-onset parkinsonism and catatonic episodes. *Parkinsonism and Related Disorders*, **2021**, 84, 5-7	3.6	
70	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. *Brain*, **2021**, 144, 3020-3035	11.2	1
69	Broadening the phenotypic spectrum of Beta3GalT6-associated phenotypes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3153-3160	2.5	0
68	CD28.OX40 co-stimulatory combination is associated with long in vivo persistence and high activity of CAR.CD30 T-cells. *Haematologica*, **2021**, 106, 987-999	6.6	16
67	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. *American Journal of Human Genetics*, **2021**, 108, 115-133	11	8
66	Melanotic Neuroectodermal Tumor of Infancy (MNTI) and Pineal Anlage Tumor (PAT) Harbor A Medulloblastoma Signature by DNA Methylation Profiling. *Cancers*, **2021**, 13,	6.6	4
65	Expanding the spectrum of EWSR1-PATZ1 rearranged CNS tumors: An infantile case with leptomeningeal dissemination. *Brain Pathology*, **2021**, 31, e12934	6	4
64	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. *Clinical Epigenetics*, **2021**, 13, 157	7.7	0
63	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in. *Genes*, **2021**, 12,	4.2	0
62	DICER1-associated malignancies mimicking germ cell neoplasms: Report of two cases and review of the literature. *Pathology Research and Practice*, **2021**, 225, 153553	3.4	3
61	Refinement of the clinical and mutational spectrum of UBE2A deficiency syndrome. *Clinical Genetics*, **2020**, 98, 172-178	4	2
60	Developmental and epileptic encephalopathy due to SZT2 genomic variants: Emerging features of a syndromic condition. *Epilepsy and Behavior*, **2020**, 108, 107097	3.2	2
59	Co-occurrence of mutations in KIF7 and KIAA0556 in Joubert syndrome with ocular coloboma, pituitary malformation and growth hormone deficiency: a case report and literature review. *BMC Pediatrics*, **2020**, 20, 120	2.6	3

58	DNA Methylation Profiling for Diagnosing Undifferentiated Sarcoma with Capicua Transcriptional Receptor () Alterations. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	10
57	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2020**, 106, 356-370	11	51
56	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. *Parkinsonism and Related Disorders*, **2020**, 72, 75-79	3.6	9
55	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. *American Journal of Human Genetics*, **2020**, 106, 484-495	11	16
54	MODL-23. DNA METHYLATION AND COPY NUMBER VARIATION PROFILE FOR CHARACTERIZATION OF PEDIATRIC BRAIN TUMOR PRIMARY CELL LINES. *Neuro-Oncology*, **2020**, 22, iii415-iii415	1	78
53	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. *American Journal of Human Genetics*, **2020**, 107, 1129-1148	11	11
52	Modeling medulloblastoma in vivo and with human cerebellar organoids. *Nature Communications*, **2020**, 11, 583	17.4	54
51	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
50	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. *Pediatric Neurology*, **2020**, 104, 40-45	2.9	12
49	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
48	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Cl/H-Exchanger, Causes Early-Onset Neurodegeneration. *American Journal of Human Genetics*, **2020**, 107, 1062-1077	11	7
47	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
46	Functional analysis of variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. *Journal of Medical Genetics*, **2020**,	5.8	2
45	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. *American Journal of Human Genetics*, **2019**, 105, 493-508	11	30
44	Expanding the clinical spectrum associated with PACS2 mutations. *Clinical Genetics*, **2019**, 95, 525-531	4	10
43	Role of DNA Methylation Profile in Diagnosing Astroblastoma: A Case Report and Literature Review. *Frontiers in Genetics*, **2019**, 10, 391	4.5	14
42	POGZ-related epilepsy: Case report and review of the literature. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1631-1636	2.5	10
41	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. *Human Mutation*, **2019**, 40, 721-728	4.7	15

40	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. *Cerebellum*, **2019**, 18, 422-432	4.3	7
39	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. *American Journal of Human Genetics*, **2019**, 105, 640-657	11	16
38	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. *Journal of Experimental Medicine*, **2019**, 216, 2778-2799	16.6	71
37	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. *Cell Death and Disease*, **2019**, 10, 201	9.8	61
36	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. *European Journal of Medical Genetics*, **2019**, 62, 103534	2.6	6
35	Functional evaluation of natural killer cell cytotoxic activity in NFKB2-mutated patients. *Immunology Letters*, **2018**, 194, 40-43	4.1	9
34	Expanding the histopathological spectrum of CFL2-related myopathies. *Clinical Genetics*, **2018**, 93, 1234-1239	4	7
33	The impact of next-generation sequencing on the diagnosis of pediatric-onset hereditary spastic paraplegias: new genotype-phenotype correlations for rare HSP-related genes. *Neurogenetics*, **2018**, 19, 111-121	3	28
32	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. *Archives of Oral Biology*, **2018**, 91, 96-102	2.8	3
31	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. *Human Molecular Genetics*, **2018**, 27, 1892-1904	5.6	18
30	VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. *BMC Bioinformatics*, **2018**, 19, 477	3.6	9
29	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2018**, 103, 621-630	11	45
28	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. *Journal of Medical Genetics*, **2018**, 55, 837-846	5.8	31
27	Biallelic mutations in early-onset, variably progressive neurodegeneration. *Neurology*, **2018**, 91, e319-e330	8.3	26
26	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. *Brain*, **2017**, 140, e34	11.2	8
25	Integrin β7 Is a Functional Marker and Potential Therapeutic Target in Glioblastoma. *Cell Stem Cell*, **2017**, 21, 35-50.e9	18	66
24	Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. *European Journal of Paediatric Neurology*, **2017**, 21, 475-484	3.8	6
23	Novel - Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. *Cancer Research*, **2017**, 77, 5860-5872	10.1	14

22	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". *American Journal of Human Genetics*, **2017**, 101, 815-823	11	22
21	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. *Orphanet Journal of Rare Diseases*, **2017**, 12, 89	4.2	28
20	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proof-of-concept examples. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1772-9	2.5	18
19	Biallelic Mutations in TBCD, Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 962-973	11	55
18	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. *American Journal of Human Genetics*, **2016**, 99, 974-983	11	37
17	Protracted late infantile ceroid lipofuscinosis due to TPP1 mutations: Clinical, molecular and biochemical characterization in three sibs. *Journal of the Neurological Sciences*, **2015**, 356, 65-71	3.2	21
16	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. *American Journal of Human Genetics*, **2015**, 96, 816-25	11	75
15	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. *Nature Genetics*, **2015**, 47, 661-7	36.3	128
14	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. *Human Mutation*, **2015**, 36, 1080-7	4.7	51
13	The emerging role of MicroRNA in schizophrenia. *CNS and Neurological Disorders - Drug Targets*, **2015**, 14, 208-21	2.6	26
12	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. *Journal of Neurology*, **2014**, 261, 870-6	5.5	41
11	Mutations in ZBTB20 cause Primrose syndrome. *Nature Genetics*, **2014**, 46, 815-7	36.3	61
10	Dynamics of the shade-avoidance response in Arabidopsis. *Plant Physiology*, **2013**, 163, 331-53	6.6	61
9	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. *Journal of Medical Genetics*, **2013**, 50, 493-9	5.8	33
8	Plant adaptation to dynamically changing environment: the shade avoidance response. *Biotechnology Advances*, **2012**, 30, 1047-58	17.8	106
7	A restricted spectrum of mutations in the SMAD4 tumor-suppressor gene underlies Myhre syndrome. *American Journal of Human Genetics*, **2012**, 90, 161-9	11	64
6	Identification of a novel cis-regulatory element for UV-B-induced transcription in Arabidopsis. *Plant Journal*, **2008**, 54, 402-14	6.9	47
5	A novel regulatory circuit underlying plant response to canopy shade. *Plant Signaling and Behavior*, **2008**, 3, 137-9	2.5	7

4	The Arabidopsis homeodomain-leucine zipper II gene family: diversity and redundancy. *Plant Molecular Biology*, **2008**, 68, 465-78	4.6	97
3	Canopy shade causes a rapid and transient arrest in leaf development through auxin-induced cytokinin oxidase activity. *Genes and Development*, **2007**, 21, 1863-8	12.6	150
2	A dynamic balance between gene activation and repression regulates the shade avoidance response in Arabidopsis. *Genes and Development*, **2005**, 19, 2811-5	12.6	192
1	ACE2gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population		25