Andrea Ciolfi

List of Publications by Year
in descending order

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80	3,637	136885	149623
papers	citations	32 h-index	56 g-index

84	84	84	7630
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A dynamic balance between gene activation and repression regulates the shade avoidance response in Arabidopsis. Genes and Development, 2005, 19, 2811-2815.	2.7	224
2	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
3	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	9.4	177
4	Canopy shade causes a rapid and transient arrest in leaf development through auxin-induced cytokinin oxidase activity. Genes and Development, 2007, 21, 1863-1868.	2.7	174
5	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	2.6	171
6	Plant adaptation to dynamically changing environment: The shade avoidance response. Biotechnology Advances, 2012, 30, 1047-1058.	6.0	155
7	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
8	The Arabidopsis Homeodomain-leucine Zipper II gene family: diversity and redundancy. Plant Molecular Biology, 2008, 68, 465-478.	2.0	112
9	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. Cell Death and Disease, 2019, 10, 201.	2.7	105
10	Modeling medulloblastoma in vivo and with human cerebellar organoids. Nature Communications, 2020, 11, 583.	5.8	105
11	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, 816-825.	2.6	102
12	Integrin Î±7 Is a Functional Marker and Potential Therapeutic Target in Glioblastoma. Cell Stem Cell, 2017, 21, 35-50.e9.	5.2	101
13	Dynamics of the Shade-Avoidance Response in Arabidopsis. Plant Physiology, 2013, 163, 331-353.	2.3	84
14	Mutations in ZBTB20 cause Primrose syndrome. Nature Genetics, 2014, 46, 815-817.	9.4	79
15	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. American Journal of Human Genetics, 2012, 90, 161-169.	2.6	77
16	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	2.6	73
17	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67
18	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.	2.6	66

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19	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. Journal of Neurology, 2014, 261, 870-876.	1.8	56
20	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.	0.7	52
21	Identification of a novel cis-regulatory element for UV-B-induced transcription in Arabidopsis. Plant Journal, 2008, 54, 402-414.	2.8	51
22	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	2.6	49
23	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.	2.6	48
24	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
25	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
26	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	1.5	44
27	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.	1.7	42
28	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
29	The Emerging Role of MicroRNA in Schizophrenia. CNS and Neurological Disorders - Drug Targets, 2015, 14, 208-221.	0.8	41
30	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. Journal of Medical Genetics, 2013, 50, 493-499.	1.5	40
31	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40
32	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. Orphanet Journal of Rare Diseases, 2017, 12, 89.	1.2	39
33	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. American Journal of Human Genetics, 2020, 107, 1129-1148.	2.6	38
34	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€• American Journal of Human Genetics, 2017, 101, 815-823.	2.6	37
35	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.	2.6	37
36	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.5	35

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37	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.	2.6	31
38	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Human Molecular Genetics, 2018, 27, 1892-1904.	1.4	29
39	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.	0.3	27
40	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proofâ€ofâ€concept examples. American Journal of Medical Genetics, Part A, 2016, 170, 1772-1779.	0.7	26
41	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotypeâ€“phenotype correlations. Human Mutation, 2019, 40, 721-728.	1.1	26
42	Role of DNA Methylation Profile in Diagnosing Astroblastoma: A Case Report and Literature Review. Frontiers in Genetics, 2019, 10, 391.	1.1	25
43	DNA Methylation Profiling for Diagnosing Undifferentiated Sarcoma with Capicua Transcriptional Receptor (CIC) Alterations. International Journal of Molecular Sciences, 2020, 21, 1818.	1.8	24
44	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Clâ¯/H+-Exchanger, Causes Early-Onset Neurodegeneration. American Journal of Human Genetics, 2020, 107, 1062-1077.	2.6	23
45	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.	1.2	23
46	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	2.6	23
47	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.	2.6	22
48	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	1.8	22
49	Novel SEC61Gâ€“EGFR Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. Cancer Research, 2017, 77, 5860-5872.	0.4	21
50	POGZâ€related epilepsy: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1631-1636.	0.7	19
51	Expanding the clinical spectrum associated with PACS2 mutations. Clinical Genetics, 2019, 95, 525-531.	1.0	18
52	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. Brain, 2017, 140, e34-e34.	3.7	17
53	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	1.0	17
54	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. European Journal of Medical Genetics, 2019, 62, 103534.	0.7	16

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55	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. Parkinsonism and Related Disorders, 2020, 72, 75-79.	1.1	16
56	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.	0.7	14
57	Developmental and epileptic encephalopathy due to SZT2 genomic variants: Emerging features of a syndromic condition. Epilepsy and Behavior, 2020, 108, 107097.	0.9	14
58	Functional evaluation of natural killer cell cytotoxic activity in NFKB2-mutated patients. Immunology Letters, 2018, 194, 40-43.	1.1	12
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.	0.7	12
60	Melanotic Neuroectodermal Tumor of Infancy (MNTI) and Pineal Anlage Tumor (PAT) Harbor A Medulloblastoma Signature by DNA Methylation Profiling. Cancers, 2021, 13, 706.	1.7	12
61	DICER1-associated malignancies mimicking germ cell neoplasms: Report of two cases and review of the literature. Pathology Research and Practice, 2021, 225, 153553.	1.0	12
62	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	1.0	11
63	Expanding the spectrum of EWSR1â€PATZ1 rearranged CNS tumors: An infantile case with leptomeningeal dissemination. Brain Pathology, 2021, 31, e12934.	2.1	11
64	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. Brain, 2021, 144, 3020-3035.	3.7	11
65	Neonatal Manifestations of Chronic Granulomatous Disease: MAS/HLH and Necrotizing Pneumonia as Unusual Phenotypes and Review of the Literature. Journal of Clinical Immunology, 2022, 42, 299-311.	2.0	11
66	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. Cerebellum, 2019, 18, 422-432.	1.4	10
67	Functional analysis of TLK2 variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. Journal of Medical Genetics, 2022, 59, 170-179.	1.5	9
68	A novel regulatory circuit underlying plant response to canopy shade. Plant Signaling and Behavior, 2008, 3, 137-139.	1.2	8
69	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. Archives of Oral Biology, 2018, 91, 96-102.	0.8	8
70	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	1.8	8
71	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	1.0	5
72	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3153-3160.	0.7	3

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73	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in LTBP3. Genes, 2021, 12, 1406.	1.0	2
74	FRI0540â€…A NOVEL AUTOINFLAMMATORY DISEASE CHARACTERIZED BY NEONATAL-ONSET CYTOPENIA WITH AUTOINFLAMMATION, RASH, AND HEMOPHAGOCYTOSIS (NOCARH) DUE TO ABERRANT CDC42 FUNCTION. , 2019, , .		1
75	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.	1.1	1
76	Regulatory networks for the shade avoidance response. Comparative Biochemistry and Physiology Part A, Molecular & Integrative Physiology, 2009, 153, S206.	0.8	0
77	Molecular mechanisms of plant adaptation to canopy shade. Journal of Biotechnology, 2010, 150, 112-113.	1.9	0
78	Cover Image, Volume 170A, Number 7, July 2016. , 2016, 170, i-i.		0
79	FRI0539â€…WNT6 MUTATION CAUSES AN EARLY ONSET GRANULOMATOSUS INTESTINAL DISEASE WITH RECURRENT HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS (HLH). , 2019, , .		0
80	MODL-23. DNA METHYLATION AND COPY NUMBER VARIATION PROFILE FOR CHARACTERIZATION OF PEDIATRIC BRAIN TUMOR PRIMARY CELL LINES. Neuro-Oncology, 2020, 22, iii415-iii415.	0.6	0