Andrew P Jackson

List of Publications by Year
in descending order

112	17,701	23544	23514
papers	citations	58 h-index	111 g-index

125	125	125	25371
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Cerebral organoids model human brain development and microcephaly. Nature, 2013, 501, 373-379.	13.7	3,889
2	Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438.	13.7	1,211
3	cGAS surveillance of micronuclei links genome instability to innate immunity. Nature, 2017, 548, 461-465.	13.7	1,158
4	Mutations in the gene encoding the 3â€²-5â€² DNA exonuclease TREX1 cause Aicardi-GoutiÃ¨res syndrome at the AGS1 locus. Nature Genetics, 2006, 38, 917-920.	9.4	752
5	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	6.5	698
6	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃ¨res syndrome and mimic congenital viral brain infection. Nature Genetics, 2006, 38, 910-916.	9.4	592
7	Mutation in the gene encoding ferritin light polypeptide causes dominant adult-onset basal ganglia disease. Nature Genetics, 2001, 28, 350-354.	9.4	533
8	Identification of Microcephalin, a Protein Implicated in Determining the Size of the Human Brain. American Journal of Human Genetics, 2002, 71, 136-142.	2.6	499
9	Enzymatic Removal of Ribonucleotides from DNA Is Essential for Mammalian Genome Integrity and Development. Cell, 2012, 149, 1008-1022.	13.5	397
10	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
11	CRISPR screens identify genomic ribonucleotides as a source of PARP-trapping lesions. Nature, 2018, 559, 285-289.	13.7	297
12	Mutations in Microcephalin Cause Aberrant Regulation of Chromosome Condensation. American Journal of Human Genetics, 2004, 75, 261-266.	2.6	292
13	Mutations in pericentrin cause Seckel syndrome with defective ATR-dependent DNA damage signaling. Nature Genetics, 2008, 40, 232-236.	9.4	281
14	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 356-359.	9.4	219
15	Lagging-strand replication shapes the mutational landscape of the genome. Nature, 2015, 518, 502-506.	13.7	213
16	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	9.4	201
17	Ribonuclease H2 mutations induce a <scp>cGAS</scp>/<scp>STING</scp>â€dependent innate immune response. EMBO Journal, 2016, 35, 831-844.	3.5	200
18	Defective removal of ribonucleotides from DNA promotes systemic autoimmunity. Journal of Clinical Investigation, 2015, 125, 413-424.	3.9	190

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19	Mutations in ORC1, encoding the largest subunit of the origin recognition complex, cause microcephalic primordial dwarfism resembling Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 350-355.	9.4	189
20	Mechanisms and pathways of growth failure in primordial dwarfism. Genes and Development, 2011, 25, 2011-2024.	2.7	180
21	BRIT1/MCPH1 links chromatin remodelling to DNA damage response. Nature Cell Biology, 2009, 11, 865-872.	4.6	175
22	Regulation of mitotic entry by microcephalin and its overlap with ATR signalling. Nature Cell Biology, 2006, 8, 725-733.	4.6	164
23	What primary microcephaly can tell us about brain growth. Trends in Molecular Medicine, 2006, 12, 358-366.	3.5	160
24	Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. Nature Genetics, 2014, 46, 1283-1292.	9.4	156
25	Primary Autosomal Recessive Microcephaly (MCPH1) Maps to Chromosome 8p22-pter. American Journal of Human Genetics, 1998, 63, 541-546.	2.6	151
26	Ribonucleotides Misincorporated into DNA Act as Strand-Discrimination Signals in Eukaryotic Mismatch Repair. Molecular Cell, 2013, 50, 323-332.	4.5	139
27	RNA:DNA hybrids are a novel molecular pattern sensed by TLR9. EMBO Journal, 2014, 33, 542-558.	3.5	133
28	The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102 individuals and recommendations for care. Genetics in Medicine, 2016, 18, 483-493.	1.1	127
29	PCNA directs type 2 RNase H activity on DNA replication and repair substrates. Nucleic Acids Research, 2011, 39, 3652-3666.	6.5	112
30	Mammalian Mitochondrial DNA Replication Intermediates Are Essentially Duplex but Contain Extensive Tracts of RNA/DNA Hybrid. Journal of Molecular Biology, 2010, 397, 1144-1155.	2.0	110
31	Gain-of-function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions. Nature Genetics, 2019, 51, 96-105.	9.4	110
32	Mutations in genes encoding condensin complex proteins cause microcephaly through decatenation failure at mitosis. Genes and Development, 2016, 30, 2158-2172.	2.7	106
33	A Third Novel Locus for Primary Autosomal Recessive Microcephaly Maps to Chromosome 9q34. American Journal of Human Genetics, 2000, 66, 724-727.	2.6	105
34	cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing. Nature Genetics, 2020, 52, 1364-1372.	9.4	105
35	The second locus for autosomal recessive primary microcephaly (MCPH2) maps to chromosome 19q13.1â€“13.2. European Journal of Human Genetics, 1999, 7, 815-820.	1.4	103
36	A Fifth Locus for Primary Autosomal Recessive Microcephaly Maps to Chromosome 1q31. American Journal of Human Genetics, 2000, 67, 1578-1580.	2.6	101

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37	Type I interferon causes thrombotic microangiopathy by a dose-dependent toxic effect on the microvasculature. Blood, 2016, 128, 2824-2833.	0.6	97
38	Meierâ€“Gorlin syndrome genotypeâ€“phenotype studies: 35 individuals with pre-replication complex gene mutations and 10 without molecular diagnosis. European Journal of Human Genetics, 2012, 20, 598-606.	1.4	95
39	The Structure of the Human RNase H2 Complex Defines Key Interaction Interfaces Relevant to Enzyme Function and Human Disease. Journal of Biological Chemistry, 2011, 286, 10530-10539.	1.6	94
40	Cree encephalitis is allelic with Aicardi-Goutieres syndrome: implications for the pathogenesis of disorders of interferon alpha metabolism. Journal of Medical Genetics, 2003, 40, 183-187.	1.5	93
41	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. American Journal of Human Genetics, 2016, 99, 125-138.	2.6	92
42	Thrombotic Microangiopathy Associated with Interferon Beta. New England Journal of Medicine, 2014, 370, 1270-1271.	13.9	89
43	Autozygosity Mapping of a Seckel Syndrome Locus to Chromosome 3q22.1-q24. American Journal of Human Genetics, 2000, 67, 498-503.	2.6	88
44	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	9.4	81
45	Evolution of primary microcephaly genes and the enlargement of primate brains. Current Opinion in Genetics and Development, 2005, 15, 241-248.	1.5	78
46	Aicardi-GoutiÃ¨res Syndrome Displays Genetic Heterogeneity with One Locus (AGS1) on Chromosome 3p21. American Journal of Human Genetics, 2000, 67, 213-221.	2.6	77
47	A Unique Set of Centrosome Proteins Requires Pericentrin for Spindle-Pole Localization and Spindle Orientation. Current Biology, 2014, 24, 2327-2334.	1.8	77
48	Hypomorphic PCNA mutation underlies a human DNA repair disorder. Journal of Clinical Investigation, 2014, 124, 3137-3146.	3.9	77
49	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	1.5	75
50	Quantifying single nucleotide variant detection sensitivity in exome sequencing. BMC Bioinformatics, 2013, 14, 195.	1.2	74
51	Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. Human Mutation, 2014, 35, 76-85.	1.1	74
52	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. Nature Genetics, 2016, 48, 36-43.	9.4	74
53	Mutations in the NHEJ Component XRCC4 Cause Primordial Dwarfism. American Journal of Human Genetics, 2015, 96, 412-424.	2.6	71
54	DNA Polymerase Epsilon Deficiency Causes IMAGe Syndrome with Variable Immunodeficiency. American Journal of Human Genetics, 2018, 103, 1038-1044.	2.6	71

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55	PolÎµ Instability Drives Replication Stress, Abnormal Development, and Tumorigenesis. Molecular Cell, 2018, 70, 707-721.e7.	4.5	69
56	Reduction of hRNase H2 activity in Aicardiâ€“GoutiÃ¨res syndrome cells leads to replication stress and genome instability. Human Molecular Genetics, 2015, 24, 649-658.	1.4	67
57	RNase H2, mutated in Aicardiâ€GoutiÃ¨res syndrome, promotes LINEâ€1 retrotransposition. EMBO Journal, 2018, 37, .	3.5	67
58	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	2.6	65
59	ANO5Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. Human Mutation, 2013, 34, 1111-1118.	1.1	64
60	Meier-Gorlin syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 114.	1.2	62
61	Genetic Defects in Human Pericentrin Are Associated With Severe Insulin Resistance and Diabetes. Diabetes, 2011, 60, 925-935.	0.3	61
62	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. American Journal of Human Genetics, 2018, 103, 553-567.	2.6	58
63	The first missense alteration in the MCPH1 gene causes autosomal recessive microcephaly with an extremely mild cellular and clinical phenotype. Human Mutation, 2005, 26, 496-496.	1.1	53
64	The presence of multiple regions of homozygous deletion at the CSMD1 locus in oral squamous cell carcinoma question the role of CSMD1 in head and neck carcinogenesis. Genes Chromosomes and Cancer, 2003, 37, 132-140.	1.5	47
65	Microcephalic Osteodysplastic Primordial Dwarfism, Type II: a Clinical Review. Current Osteoporosis Reports, 2017, 15, 61-69.	1.5	47
66	The expanding phenotype of RNU4ATAC pathogenic variants to Lowry Wood syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 465-469.	0.7	45
67	Microcephalin: A Causal Link Between Impaired Damage Response Signalling and Microcephaly. Cell Cycle, 2006, 5, 2339-2344.	1.3	44
68	Meierâ€“Gorlin syndrome: Growth and secondary sexual development of a microcephalic primordial dwarfism disorder. American Journal of Medical Genetics, Part A, 2012, 158A, 2733-2742.	0.7	44
69	Type I interferon dysregulation and neurological disease. Nature Reviews Neurology, 2015, 11, 515-523.	4.9	43
70	A gene for ataxic cerebral palsy maps to chromosome 9p12â€“q12. European Journal of Human Genetics, 2000, 8, 267-272.	1.4	42
71	Genome-wide mapping of embedded ribonucleotides and other noncanonical nucleotides using emRiboSeq and EndoSeq. Nature Protocols, 2015, 10, 1433-1444.	5.5	42
72	Condensin II mutation causes T-cell lymphoma through tissue-specific genome instability. Genes and Development, 2016, 30, 2173-2186.	2.7	41

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73	Growth in individuals with Majewski osteodysplastic primordial dwarfism type II caused by pericentrin mutations. American Journal of Medical Genetics, Part A, 2012, 158A, 2719-2725.	0.7	40
74	Ribonucleotide Excision Repair Is Essential to Prevent Squamous Cell Carcinoma of the Skin. Cancer Research, 2018, 78, 5917-5926.	0.4	40
75	Microcephalin coordinates mitosis in the syncytialDrosophilaembryo. Journal of Cell Science, 2007, 120, 3578-3588.	1.2	39
76	Signatures of TOP1 transcription-associated mutagenesis in cancer and germline. Nature, 2022, 602, 623-631.	13.7	38
77	Ribonuclease H2 in health and disease. Biochemical Society Transactions, 2014, 42, 717-725.	1.6	37
78	Heterozygous lamin B1 and lamin B2 variants cause primary microcephaly and define a novel laminopathy. Genetics in Medicine, 2021, 23, 408-414.	1.1	35
79	Nucleic acidâ€mediated inflammatory diseases. BioEssays, 2008, 30, 833-842.	1.2	34
80	A novel nonsense CDK5RAP2 mutation in a Somali child with primary microcephaly and sensorineural hearing loss. American Journal of Medical Genetics, Part A, 2012, 158A, 2577-2582.	0.7	34
81	A second locus for Aicardi-Goutieres syndrome at chromosome 13q14-21. Journal of Medical Genetics, 2005, 43, 444-450.	1.5	33
82	A sensitive and affordable multiplex RT-qPCR assay for SARS-CoV-2 detection. PLoS Biology, 2020, 18, e3001030.	2.6	32
83	SET Nuclear Oncogene Associates with Microcephalin/MCPH1 and Regulates Chromosome Condensation. Journal of Biological Chemistry, 2011, 286, 21393-21400.	1.6	30
84	Replication Proteins and Human Disease. Cold Spring Harbor Perspectives in Biology, 2014, 6, a013060-a013060.	2.3	30
85	Low-frequency variation in TP53 has large effects on head circumference and intracranial volume. Nature Communications, 2019, 10, 357.	5.8	30
86	Linked-read genome sequencing identifies biallelic pathogenic variants in DONSON as a novel cause of Meier-Gorlin syndrome. Journal of Medical Genetics, 2020, 57, 195-202.	1.5	29
87	Trisomy 12 mosaicism in a 7 year old girl with dysmorphic features and normal mental development.. Journal of Medical Genetics, 1994, 31, 253-254.	1.5	28
88	17q21.31 Microdeletion Syndrome: Further Expanding the Clinical Phenotype. Cytogenetic and Genome Research, 2009, 127, 61-66.	0.6	27
89	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
90	DONSON and FANCM associate with different replisomes distinguished by replication timing and chromatin domain. Nature Communications, 2020, 11, 3951.	5.8	26

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91	ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. PLoS Genetics, 2019, 15, e1007605.	1.5	25
92	A new locus for autosomal recessive non-syndromal sensorineural hearing impairment (DFNB27) on chromosome 2q23â€“q31. European Journal of Human Genetics, 2000, 8, 991-993.	1.4	21
93	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98.	1.7	20
94	PRIM1 deficiency causes a distinctive primordial dwarfism syndrome. Genes and Development, 2020, 34, 1520-1533.	2.7	20
95	The continuing failure to recognise Alstrom syndrome and further evidence of genetic homogeneity. Journal of Medical Genetics, 2000, 37, 219-219.	1.5	16
96	An autosomal recessive exfoliative ichthyosis with linkage to chromosome 12q13. British Journal of Dermatology, 2003, 149, 174-180.	1.4	16
97	Synonymous Mutations inRNASEH2ACreate Cryptic Splice Sites Impairing RNase H2 Enzyme Function in Aicardi-GoutiÃ¨res Syndrome. Human Mutation, 2013, 34, 1066-1070.	1.1	16
98	Biallelic variants in DNA2 cause microcephalic primordial dwarfism. Human Mutation, 2019, 40, 1063-1070.	1.1	16
99	Sonic hedgehog accelerates DNA replication to cause replication stress promoting cancer initiation in medulloblastoma. Nature Cancer, 2020, 1, 840-854.	5.7	16
100	Two novel mutations in RNU4ATAC in two siblings with an atypical mild phenotype of microcephalic osteodysplastic primordial dwarfism type 1. Clinical Dysmorphology, 2016, 25, 68-72.	0.1	12
101	Defining the clinical phenotype of Saulâ€“Wilson syndrome. Genetics in Medicine, 2020, 22, 857-866.	1.1	11
102	Mental retardation, keratoconus, febrile seizures and sinoatrial block: a previously undescribed autosomal recessive disorder. Clinical Genetics, 2005, 67, 448-449.	1.0	10
103	Analysis of novel missense ATR mutations reveals new splicing defects underlying Seckel syndrome. Human Mutation, 2018, 39, 1847-1853.	1.1	10
104	Identification of Small-Molecule Inhibitors of the Ribonuclease H2 Enzyme. Journal of Biomolecular Screening, 2013, 18, 610-620.	2.6	8
105	Rare variants of the 3â€™-5â€™ DNA exonuclease TREX1 in early onset small vessel stroke. Wellcome Open Research, 2017, 2, 106.	0.9	7
106	Diversifying microtubules in brain development. Nature Genetics, 2009, 41, 638-640.	9.4	4
107	Growth in individuals with Saulâ€“Wilson syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2110-2116.	0.7	4
108	Exploring microcephaly and human brain evolution. Developmental Medicine and Child Neurology, 2012, 54, 580-581.	1.1	3

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109	This should not be the end for terminator technology in GM crops. Nature, 1999, 402, 457-457.	13.7	2
110	Simultaneous Intestinal and Kidney Transplantation in Adults. Journal of Investigative Surgery, 2019, 32, 283-289.	0.6	2
111	Murray and Jackson reply. Developmental Medicine and Child Neurology, 2013, 55, 194-194.	1.1	0
112	A Unique Set of Centrosome Proteins Requires Pericentrin for Spindle-Pole Localization and Spindle Orientation. Current Biology, 2014, 24, 2975.	1.8	0