Michael A Simpson

List of Publications by Year
in descending order

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190 papers	13,147 citations	18482 62 h-index	26613 107 g-index
196 all docs	196 docs citations	196 times ranked	24366 citing authors

#	Article	IF	Citations
1	Assessment of Minimal Residual Disease in Standard-Risk AML. New England Journal of Medicine, 2016, 374, 422-433.	27.0	662
2	Mutations in IL36RN/IL1F5 Are Associated with the Severe Episodic Inflammatory Skin Disease Known as Generalized Pustular Psoriasis. American Journal of Human Genetics, 2011, 89, 432-437.	6.2	468
3	Mutations in GATA2 cause primary lymphedema associated with a predisposition to acute myeloid leukemia (Emberger syndrome). Nature Genetics, 2011, 43, 929-931.	21.4	440
4	Infantile-onset symptomatic epilepsy syndrome caused by a homozygous loss-of-function mutation of GM3 synthase. Nature Genetics, 2004, 36, 1225-1229.	21.4	359
5	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
6	Mutations in NOTCH2 cause Hajdu-Cheney syndrome, a disorder of severe and progressive bone loss. Nature Genetics, 2011, 43, 303-305.	21.4	291
7	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. Nature Genetics, 2013, 45, 1244-1248.	21.4	289
8	Novel Mutation in Desmoplakin Causes Arrhythmogenic Left Ventricular Cardiomyopathy. Circulation, 2005, 112, 636-642.	1.6	266
9	Deficiency of terminal ADP-ribose protein glycohydrolase TARG1/C6orf130 in neurodegenerative disease. EMBO Journal, 2013, 32, 1225-1237.	7.8	263
10	Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. Nature Genetics, 2013, 45, 1300-1308.	21.4	247
11	Novel mutations in PIEZO1 cause an autosomal recessive generalized lymphatic dysplasia with non-immune hydrops fetalis. Nature Communications, 2015, 6, 8085.	12.8	247
12	Mutations in TJP2 cause progressive cholestatic liver disease. Nature Genetics, 2014, 46, 326-328.	21.4	244
13	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601.	12.8	233
14	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	21.4	231
15	De Novo Mutations in MLL Cause Wiedemann-Steiner Syndrome. American Journal of Human Genetics, 2012, 91, 358-364.	6.2	225
16	Genomic and clinical profiling of a national nephrotic syndrome cohort advocates a precision medicine approach to disease management. Kidney International, 2017, 91, 937-947.	5.2	201
17	Mutations in FAM20C Are Associated with Lethal Osteosclerotic Bone Dysplasia (Raine Syndrome), Highlighting a Crucial Molecule in Bone Development. American Journal of Human Genetics, 2007, 81, 906-912.	6.2	190
18	Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. Nature, 2013, 498, 232-235.	27.8	184

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19	Sequence Alterations within CYP7B1 Implicate Defective Cholesterol Homeostasis in Motor-Neuron Degeneration. American Journal of Human Genetics, 2008, 82, 510-515.	6.2	171
20	Mutations in ZMYND10, a Gene Essential for Proper Axonemal Assembly of Inner and Outer Dynein Arms in Humans and Flies, Cause Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 346-356.	6.2	167
21	Maspardin Is Mutated in Mast Syndrome, a Complicated Form of Hereditary Spastic Paraplegia Associated with Dementia. American Journal of Human Genetics, 2003, 73, 1147-1156.	6.2	158
22	AP1S3 Mutations Are Associated with Pustular Psoriasis and Impaired Toll-like Receptor 3 Trafficking. American Journal of Human Genetics, 2014, 94, 790-797.	6.2	153
23	Germline FH Mutations Presenting With Pheochromocytoma. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2046-E2050.	3.6	147
24	Mutation in Vascular Endothelial Growth Factor-C, a Ligand for Vascular Endothelial Growth Factor Receptor-3, Is Associated With Autosomal Dominant Milroy-Like Primary Lymphedema. Circulation Research, 2013, 112, 956-960.	4.5	143
25	Rare Pathogenic Variants in IL36RN Underlie a Spectrum of Psoriasis-Associated Pustular Phenotypes. Journal of Investigative Dermatology, 2013, 133, 1366-1369.	0.7	140
26	Sequencing of human genomes with nanopore technology. Nature Communications, 2019, 10, 1869.	12.8	140
27	Mutations in KIF11 Cause Autosomal-Dominant Microcephaly Variably Associated with Congenital Lymphedema and Chorioretinopathy. American Journal of Human Genetics, 2012, 90, 356-362.	6.2	138
28	Î³-Secretase Mutations in Hidradenitis Suppurativa: New Insights into Disease Pathogenesis. Journal of Investigative Dermatology, 2013, 133, 601-607.	0.7	133
29	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
30	HLA-C*06:02 genotype is a predictive biomarker of biologic treatment response in psoriasis. Journal of Allergy and Clinical Immunology, 2019, 143, 2120-2130.	2.9	128
31	Mutations in the Î³-Secretase Genes NCSTN , PSENEN , and PSEN1 Underlie Rare Forms of Hidradenitis Suppurativa (Acne Inversa). Journal of Investigative Dermatology, 2012, 132, 2459-2461.	0.7	126
32	Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. Haematologica, 2016, 101, 1170-1179.	3.5	119
33	Enrichment of FLI1 and RUNX1 mutations in families with excessive bleeding and platelet dense granule secretion defects. Blood, 2013, 122, 4090-4093.	1.4	108
34	Rare Variations in IL36RN in Severe Adverse Drug Reactions Manifesting as Acute Generalized Exanthematous Pustulosis. Journal of Investigative Dermatology, 2013, 133, 1904-1907.	0.7	107
35	Next generation exome sequencing of paediatric inflammatory bowel disease patients identifies rare and novel variants in candidate genes. Gut, 2013, 62, 977-984.	12.1	104
36	PSENEN and NCSTN Mutations in Familial Hidradenitis Suppurativa (Acne Inversa). Journal of Investigative Dermatology, 2011, 131, 1568-1570.	0.7	103

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37	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. American Journal of Human Genetics, 2011, 88, 574-585.	6.2	100
38	Patient-specific induced-pluripotent stem cells-derived cardiomyocytes recapitulate the pathogenic phenotypes of dilated cardiomyopathy due to a novel DES mutation identified by whole exome sequencing. Human Molecular Genetics, 2013, 22, 1395-1403.	2.9	98
39	Mutations in FAM20C also identified in nonâ€lethal osteosclerotic bone dysplasia. Clinical Genetics, 2009, 75, 271-276.	2.0	97
40	Rapid identification of mutations in GJC2 in primary lymphoedema using whole exome sequencing combined with linkage analysis with delineation of the phenotype. Journal of Medical Genetics, 2011, 48, 251-255.	3.2	96
41	Candidate driver genes involved in genome maintenance and DNA repair in SÃ©zary syndrome. Blood, 2016, 127, 3387-3397.	1.4	96
42	Mitochondria function associated genes contribute to Parkinsonâ€™s Disease risk and later age at onset. Npj Parkinson's Disease, 2019, 5, 8.	5.3	95
43	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
44	Predicting the functional consequences of non-synonymous DNA sequence variants â€” evaluation of bioinformatics tools and development of a consensus strategy. Genomics, 2013, 102, 223-228.	2.9	89
45	Germline Mutations in the CDKN2B Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. Cancer Discovery, 2015, 5, 723-729.	9.4	88
46	Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome. American Journal of Human Genetics, 2015, 96, 153-161.	6.2	88
47	De Novo Mutations of the Gene Encoding the Histone Acetyltransferase KAT6B Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 290-294.	6.2	86
48	Homozygous Mutation of Desmocollin-2 in Arrhythmogenic Right Ventricular Cardiomyopathy with Mild Palmoplantar Keratoderma and Woolly Hair. Cardiology, 2009, 113, 28-34.	1.4	85
49	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	2.0	85
50	EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088.	8.2	83
51	Genome-wide association study in frontal fibrosing alopecia identifies four susceptibility loci including HLA-B*07:02. Nature Communications, 2019, 10, 1150.	12.8	82
52	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	7.6	81
53	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
54	Characterization of the clinical and immunologic phenotype and management of 157 individuals with 56 distinct heterozygous NFKB1 mutations. Journal of Allergy and Clinical Immunology, 2020, 146, 901-911.	2.9	78

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55	Defective Mitochondrial mRNA Maturation Is Associated with Spastic Ataxia. American Journal of Human Genetics, 2010, 87, 655-660.	6.2	76
56	Recurrent heterozygous missense mutation, p.Gly573Ser, in the TRPV3 gene in an Indian boy with sporadic Olmsted syndrome. British Journal of Dermatology, 2012, 167, 440-442.	1.5	75
57	A Three-Stage Genome-Wide Association Study of General Cognitive Ability: Hunting the Small Effects. Behavior Genetics, 2010, 40, 759-767.	2.1	74
58	Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. British Journal of Dermatology, 2015, 172, 94-100.	1.5	74
59	Epithelial Inflammation Resulting from an Inherited Loss-of-Function Mutation in EGFR. Journal of Investigative Dermatology, 2014, 134, 2570-2578.	0.7	71
60	The role of <scp>SLC</scp>2A1 mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of <scp>GLUT</scp>1 deficiency syndrome. Epilepsia, 2015, 56, e203-8.	5.1	71
61	SLFN14 mutations underlie thrombocytopenia with excessive bleeding and platelet secretion defects. Journal of Clinical Investigation, 2015, 125, 3600-3605.	8.2	71
62	Germline CDH1 mutations in bilateral lobular carcinoma in situ. British Journal of Cancer, 2014, 110, 1053-1057.	6.4	70
63	Genome-wide association study identifies three novel susceptibility loci for severe Acne vulgaris. Nature Communications, 2014, 5, 4020.	12.8	68
64	Homozygous mutation of MYBPC3 associated with severe infantile hypertrophic cardiomyopathy at high frequency among the Amish. Heart, 2007, 94, 1326-1330.	2.9	66
65	Frontal fibrosing alopecia: reflections and hypotheses on aetiology and pathogenesis. Experimental Dermatology, 2016, 25, 847-852.	2.9	66
66	Cardiac Genetic Predisposition in SuddenÂInfant Death Syndrome. Journal of the American College of Cardiology, 2018, 71, 1217-1227.	2.8	66
67	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. American Journal of Human Genetics, 2012, 91, 1115-1121.	6.2	65
68	Psoriasis and Genetics. Acta Dermato-Venereologica, 2020, 100, 55-65.	1.3	64
69	Use of nextâ€generation sequencing and candidate gene analysis to identify underlying defects in patients with inherited platelet function disorders. Journal of Thrombosis and Haemostasis, 2015, 13, 643-650.	3.8	63
70	Dysfunction of NaV1.4, a skeletal muscle voltage-gated sodium channel, in sudden infant death syndrome: a case-control study. Lancet, The, 2018, 391, 1483-1492.	13.7	63
71	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
72	Serpentine fibula polycystic kidney syndrome is part of the phenotypic spectrum of Hajduâ€“Cheney syndrome. European Journal of Human Genetics, 2012, 20, 122-124.	2.8	60

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73	Pooled Sequencing of 531 Genes in Inflammatory Bowel Disease Identifies an Associated Rare Variant in BTNL2 and Implicates Other Immune Related Genes. PLoS Genetics, 2015, 11, e1004955.	3.5	59
74	MAGI2 Mutations Cause Congenital Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2017, 28, 1614-1621.	6.1	59
75	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
76	Impact of next generation sequencing on diagnostics in a genetic skin disease clinic. Experimental Dermatology, 2013, 22, 825-831.	2.9	56
77	A genome-wide association study for extremely high intelligence. Molecular Psychiatry, 2018, 23, 1226-1232.	7.9	54
78	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in aÂSpectrum of Keratinization Disorders Associated with Thrombocytopenia. Journal of Investigative Dermatology, 2017, 137, 2344-2353.	0.7	53
79	A novel locus for an autosomal recessive hereditary spastic paraplegia (SPG35) maps to 16q21-q23. Neurology, 2008, 71, 248-252.	1.1	52
80	De novo mutations implicate novel genes in systemic lupus erythematosus. Human Molecular Genetics, 2018, 27, 421-429.	2.9	52
81	A novel NIPA1 mutation associated with a pure form of autosomal dominant hereditary spastic paraplegia. Neurogenetics, 2005, 6, 79-84.	1.4	51
82	Cross-phenotype association mapping of the MHC identifies genetic variants that differentiate psoriatic arthritis from psoriasis. Annals of the Rheumatic Diseases, 2017, 76, 1774-1779.	0.9	51
83	Mutations in GRHL2 Result in an Autosomal-Recessive Ectodermal Dysplasia Syndrome. American Journal of Human Genetics, 2014, 95, 308-314.	6.2	48
84	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	2.8	48
85	Genome-wide meta-analysis implicates mediators of hair follicle development and morphogenesis in risk for severe acne. Nature Communications, 2018, 9, 5075.	12.8	48
86	Genotypeâ€“phenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. British Journal of Dermatology, 2020, 182, 729-737.	1.5	47
87	Mutation in GNE is associated with severe congenital thrombocytopenia. Blood, 2018, 132, 1855-1858.	1.4	46
88	An In-Depth Characterization of the Major Psoriasis Susceptibility Locus Identifies Candidate Susceptibility Alleles within an HLA-C Enhancer Element. PLoS ONE, 2013, 8, e71690.	2.5	45
89	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
90	Homozygous Missense Mutation inIL36RNin Generalized Pustular Dermatosis With Intraoral Involvement Compatible With Both AGEP and Generalized Pustular Psoriasis. JAMA Dermatology, 2015, 151, 452.	4.1	44

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91	A new locus for autosomal recessive complicated hereditary spastic paraplegia (SPG26) maps to chromosome 12p11.1-12q14. Journal of Medical Genetics, 2005, 42, 80-82.	3.2	43
92	The future of genomics for developmentalists. Development and Psychopathology, 2013, 25, 1263-1278.	2.3	41
93	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. Human Molecular Genetics, 2017, 26, 4301-4313.	2.9	41
94	Acral Peeling Skin Syndrome Resulting from a Homozygous Nonsense Mutation in the <scp>CSTA</scp> Gene Encoding Cystatin A. Pediatric Dermatology, 2013, 30, e87-8.	0.9	39
95	Genetic architecture of acne vulgaris. Journal of the European Academy of Dermatology and Venereology, 2017, 31, 1978-1990.	2.4	39
96	Epistemic uncertainties and natural hazard risk assessment â€“ PartÂ2: What should constitute good practice?. Natural Hazards and Earth System Sciences, 2018, 18, 2769-2783.	3.6	37
97	Exome Sequencing and Rare Variant Analysis RevealsÂMultiple Filaggrin Mutations in BangladeshiÂFamilies with Atopic Eczema andÂAdditional Risk Genes. Journal of Investigative Dermatology, 2018, 138, 2674-2677.	0.7	37
98	De novo single-nucleotide and copy number variation in discordant monozygotic twins reveals disease-related genes. European Journal of Human Genetics, 2019, 27, 1121-1133.	2.8	37
99	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
100	Importance of Variant Interpretation in Whole-Exome Molecular Autopsy. Circulation, 2018, 137, 2705-2715.	1.6	36
101	A mutation in NFÎºB interacting protein 1 causes cardiomyopathy and woolly haircoat syndrome of Poll Hereford cattle. Animal Genetics, 2009, 40, 42-46.	1.7	34
102	The ErbB4 CYT2 variant protects EGFR from ligand-induced degradation to enhance cancer cell motility. Science Signaling, 2014, 7, ra78.	3.6	34
103	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. American Journal of Human Genetics, 2017, 100, 364-370.	6.2	32
104	Novel mutations in the pejvakin gene are associated with autosomal recessive nonâ€syndromic hearing loss in Iranian families. Clinical Genetics, 2007, 72, 261-263.	2.0	31
105	The ADAMTS13â€“VWF axis is dysregulated in chronic thromboembolic pulmonary hypertension. European Respiratory Journal, 2019, 53, 1801805.	6.7	31
106	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	5.1	30
107	Founder mutation in dystonin-e underlying autosomal recessive epidermolysis bullosa simplex in Kuwait. British Journal of Dermatology, 2015, 172, 527-531.	1.5	29
108	Endogenous spartin, mutated in hereditary spastic paraplegia, has a complex subcellular localization suggesting diverse roles in neurons. Experimental Cell Research, 2006, 312, 2764-2777.	2.6	28

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109	Germline ESR2 mutation predisposes to medullary thyroid carcinoma and causes up-regulation of RET expression. Human Molecular Genetics, 2016, 25, 1836-1845.	2.9	28
110	MED12, TERT promoter and RBM15 mutations in primary and recurrent phyllodes tumours. British Journal of Cancer, 2018, 118, 277-284.	6.4	28
111	Thinking positively: The genetics of high intelligence. Intelligence, 2015, 48, 123-132.	3.0	27
112	De novo DNM1L mutation associated with mitochondrial epilepsy syndrome with fever sensitivity. Neurology: Genetics, 2018, 4, e258.	1.9	27
113	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
114	The Missense Mutation p.R1303Q in Type XVII Collagen Underlies Junctional Epidermolysis Bullosa Resembling Kindler Syndrome. Journal of Investigative Dermatology, 2014, 134, 845-849.	0.7	24
115	Progressive hyperpigmentation in a Taiwanese child due to an inborn error of vitamin B12 metabolism (cblJ). British Journal of Dermatology, 2015, 172, 1111-1115.	1.5	24
116	Congenital Anonychia and Uncombable Hair Syndrome: Coinheritance of Homozygous Mutations in RSPO4 and PADI3. Journal of Investigative Dermatology, 2017, 137, 1176-1179.	0.7	23
117	Pathogenic variants in HTRA2 cause an earlyâ€onset mitochondrial syndrome associated with 3â€methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2017, 40, 121-130.	3.6	23
118	Frequency of Pathogenic Germline Variants in CDH1, BRCA2, CHEK2, PALB2, BRCA1, and TP53 in Sporadic Lobular Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2019, 28, 1162-1168.	2.5	23
119	Genome-wide association meta-analysis identifies 29 new acne susceptibility loci. Nature Communications, 2022, 13, 702.	12.8	23
120	Syndromic inherited poikiloderma due to a de novo mutation in <scp>FAM</scp> 111B . British Journal of Dermatology, 2017, 176, 534-536.	1.5	22
121	Complicated Hereditary Spastic Paraplegia with Thin Corpus Callosum (HSPâ€TCC) and Childhood Onset. Neuropediatrics, 2005, 36, 274-278.	0.6	21
122	Acrocallosal syndrome: Identification of a novel KIF7 mutation and evidence for oligogenic inheritance. European Journal of Medical Genetics, 2013, 56, 39-42.	1.3	21
123	Whole-exome sequencing diagnosis of two autosomal recessive disorders in one family. British Journal of Dermatology, 2015, 172, 1407-1411.	1.5	21
124	A genome-wide analysis of putative functional and exonic variation associated with extremely high intelligence. Molecular Psychiatry, 2016, 21, 1145-1151.	7.9	20
125	Raine syndrome: a clinical, radiographic and genetic investigation of a case from the Indian subcontinent. Clinical Dysmorphology, 2010, 19, 153-156.	0.3	19
126	A combination of mutations in AKR1D1 and SKIV2L in a family with severe infantile liver disease. Orphanet Journal of Rare Diseases, 2013, 8, 74.	2.7	17

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127	Truncating mutation in intracellular phospholipase A1 gene (DDHD2) in hereditary spastic paraplegia with intellectual disability (SPG54). BMC Research Notes, 2015, 8, 271.	1.4	17
128	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. Human Mutation, 2016, 37, 1157-1161.	2.5	17
129	Novel homozygous missense mutation in NT5C2 underlying hereditary spastic paraplegia SPG45. American Journal of Medical Genetics, Part A, 2017, 173, 3109-3113.	1.2	17
130	Exome-Wide Rare Variant Analyses in Sudden Infant Death Syndrome. Journal of Pediatrics, 2018, 203, 423-428.e11.	1.8	17
131	Frequency of pathogenic germline variants in BRCA1, BRCA2, PALB2, CHEK2 and TP53 in ductal carcinoma in situ diagnosed in women under the age of 50â€‰years. Breast Cancer Research, 2019, 21, 58.	5.0	17
132	Genetic variant of TTLL11 gene and subsequent ciliary defects are associated with idiopathic scoliosis in a 5-generation UK family. Scientific Reports, 2021, 11, 11026.	3.3	16
133	Phenome-wide association study using research participantsâ€™ self-reported data provides insight into the Th17 and IL-17 pathway. PLoS ONE, 2017, 12, e0186405.	2.5	16
134	Diversity and impact of rare variants in genes encoding the platelet G protein-coupled receptors. Thrombosis and Haemostasis, 2015, 113, 826-837.	3.4	15
135	Comparative Genetic Analysis of Psoriatic Arthritis and Psoriasis for the Discovery of Genetic Risk Factors and Risk Prediction Modeling. Arthritis and Rheumatology, 2022, 74, 1535-1543.	5.6	15
136	PIK3CA mutations are common in lobular carcinoma in situ, but are not a biomarker of progression. Breast Cancer Research, 2017, 19, 7.	5.0	14
137	Homozygous acceptor splice site mutation in DSG1 disrupts plakoglobin localization and results in keratoderma and skin fragility. Journal of Dermatological Science, 2018, 89, 198-201.	1.9	14
138	Exome Sequencing and Genotyping Identify a Rare Variant inNLRP7Gene Associated With Ulcerative Colitis. Journal of Crohn's and Colitis, 2018, 12, 321-326.	1.3	14
139	Lethal Cystic Kidney Disease in Amish Neonates Associated With Homozygous Nonsense Mutation of NPHP3. American Journal of Kidney Diseases, 2009, 53, 790-795.	1.9	13
140	Rodriguez acrofacial dysostosis is caused by apparently de novo heterozygous mutations in the SF3B4 gene. American Journal of Medical Genetics, Part A, 2016, 170, 3133-3137.	1.2	13
141	Bi-allelic nonsense mutations inABHD5 underlie a mild phenotype of Dorfman-Chanarin syndrome. Journal of Dermatological Science, 2016, 81, 134-136.	1.9	13
142	Novel <scp>ADA</scp>2 mutation presenting with neutropenia, lymphopenia and bone marrow failure in patients with deficiency in adenosine deaminase 2 (<scp>DADA</scp>2). British Journal of Haematology, 2019, 186, e60-e64.	2.5	13
143	<scp>EGFR</scp> mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. Molecular Genetics & Genomic Medicine, 2015, 3, 452-458.	1.2	12
144	Semidominant GPNMB Mutations in Amyloidosis Cutis Dyschromica. Journal of Investigative Dermatology, 2019, 139, 2550-2554.e9.	0.7	12

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145	Coagulation Factor XIII-A Subunit Missense Mutation in the Pathobiology of Autosomal Dominant Multiple Dermatofibromas. Journal of Investigative Dermatology, 2020, 140, 624-635.e7.	0.7	12
146	Mutations in SPATA13/ASEF2 cause primary angle closure glaucoma. PLoS Genetics, 2020, 16, e1008721.	3.5	12
147	Next generation diagnostics of heritable connective tissue disorders. Matrix Biology, 2014, 33, 35-40.	3.6	11
148	Hidradenitis suppurativa: haploinsufficiency of gamma-secretase components does not affect gamma-secretase enzyme activityin vitro. British Journal of Dermatology, 2016, 175, 632-635.	1.5	11
149	Patients with triple-negative, JAK2V617F- and CALR-mutated essential thrombocythemia share a unique gene expression signature. Blood Advances, 2021, 5, 1059-1068.	5.2	11
150	Differences in Clinical Features and Comorbid Burden between HLA-Câ^—06:02 Carrier Groups in >9,000 People with Psoriasis. Journal of Investigative Dermatology, 2022, 142, 1617-1628.e10.	0.7	11
151	Genetic variation in schlafen genes in a patient with a recapitulation of the murine Elektra phenotype. Journal of Allergy and Clinical Immunology, 2014, 133, 1462-1465.e5.	2.9	10
152	Induced Pluripotent Stem Cell Differentiation and Three-Dimensional Tissue Formation Attenuate Clonal Epigenetic Differences in Trichohyalin. Stem Cells and Development, 2016, 25, 1366-1375.	2.1	10
153	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	1.4	10
154	Recessive Mutation in FAM83G Associated with Palmoplantar Keratoderma and Exuberant Scalp Hair. Journal of Investigative Dermatology, 2018, 138, 984-987.	0.7	10
155	Application of information theoretic feature selection and machine learning methods for the development of genetic risk prediction models. Scientific Reports, 2021, 11, 23335.	3.3	10
156	Non-syndromic severe hypodontia caused by a novel frameshift insertion mutation in the homeobox of the MSX1 gene. Archives of Oral Biology, 2017, 75, 8-13.	1.8	9
157	Noncardiac genetic predisposition in sudden infant death syndrome. Genetics in Medicine, 2019, 21, 641-649.	2.4	9
158	Confirmation of Cause of Death Via Comprehensive Autopsy and Whole Exome Molecular Sequencing in People With Epilepsy and Sudden Unexpected Death. Journal of the American Heart Association, 2021, 10, e021170.	3.7	9
159	Next-Generation Diagnostics for Genodermatoses. Journal of Investigative Dermatology, 2012, 132, E27-E28.	0.7	8
160	Identification of Rare, Disease-Associated Variants in the Promoter Region of the RNF114 Psoriasis Susceptibility Gene. Journal of Investigative Dermatology, 2012, 132, 1297-1299.	0.7	8
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