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	Differences in Clinical Features and Comorbid Burden between HLA-Cî—06:02 Carrier Groups in >9,000 People with Psoriasis. Journal of Investigative Dermatology, 2022, 142, 1617-1628.e10.	0.7	11
2	Genome-wide association meta-analysis identifies 29 new acne susceptibility loci. Nature Communications, 2022, 13, 702.	12.8	23
3	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
4	Autosomal recessive hypotrichosis with loose anagen hairs associated with TKFC mutations*. British Journal of Dermatology, 2021, 184, 935-943.	1.5	7
5	A germline mutation in the plateletâ€derived growth factor receptor beta gene may be implicated in hereditary progressive mucinous histiocytosis. British Journal of Dermatology, 2021, 184, 967-970.	1.5	7
6	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
7	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
8	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
9	Assessing BRCA1 activity in DNA damage repair using human induced pluripotent stem cells as an approach to assist classification of BRCA1 variants of uncertain significance. PLoS ONE, 2021, 16, e0260852.	2.5	2
10	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
11	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
12	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. Clinical and Experimental Dermatology, 2020, 45, 391-394.	1.3	1
13	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
14	Homozygous Nonsense Mutation in DSC3 Resulting in Skin Fragility and Hypotrichosis. Journal of Investigative Dermatology, 2020, 140, 1285-1288.	0.7	8
15	Blaschkoâ€linear lichen planus: Clinicopathological and genetic analysis. Journal of Dermatology, 2020, 47, e384-e385.	1.2	1
16	New Homozygous Missense <i>MSMO1</i> Mutation in Two Siblings with SC4MOL Deficiency Presenting with Psoriasiform Dermatitis. Cytogenetic and Genome Research, 2020, 160, 523-530.	1.1	6
17	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. Epilepsia, 2020, 61, 995-1007.	5.1	30
18	Molecular basis and inheritance patterns of amyloidosis cutis dyschromica. Clinical and Experimental Dermatology, 2020, 45, 650-653.	1.3	7

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19	Mutations in SPATA13/ASEF2 cause primary angle closure glaucoma. PLoS Genetics, 2020, 16, e1008721.	3.5	12
20	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
21	Psoriasis and Genetics. Acta Dermato-Venereologica, 2020, 100, 55-65.	1.3	64
22	Noncardiac genetic predisposition in sudden infant death syndrome. Genetics in Medicine, 2019, 21, 641-649.	2.4	9
23	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
24	Semidominant GPNMB Mutations in Amyloidosis Cutis Dyschromica. Journal of Investigative Dermatology, 2019, 139, 2550-2554.e9.	0.7	12
25	The ADAMTS13â€“VWF axis is dysregulated in chronic thromboembolic pulmonary hypertension. European Respiratory Journal, 2019, 53, 1801805.	6.7	31
26	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
27	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
28	Sequencing of human genomes with nanopore technology. Nature Communications, 2019, 10, 1869.	12.8	140
29	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
30	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
31	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
32	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
33	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
34	PADI3 , hair disorders and genomic investigation. British Journal of Dermatology, 2019, 181, 1115-1116.	1.5	1
35	Consanguinity and Double Recessive Gene Pathology. JAMA Dermatology, 2019, 155, 257.	4.1	5
36	Molecular, Epigenetic and Gene Expression Profiling of Triple Negative Essential Thrombocythaemia. Blood, 2019, 134, 308-308.	1.4	0

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37	Text-mined phenotype annotation and vector-based similarity to improve identification of similar phenotypes and causative genes in monogenic disease patients. Human Mutation, 2018, 39, 643-652.	2.5	4
38	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
39	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
40	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
41	Recessive Mutation in FAM83G Associated with Palmoplantar Keratoderma and Exuberant Scalp Hair. Journal of Investigative Dermatology, 2018, 138, 984-987.	0.7	10
42	MED12, TERT promoter and RBM15 mutations in primary and recurrent phyllodes tumours. British Journal of Cancer, 2018, 118, 277-284.	6.4	28
43	De novo mutations implicate novel genes in systemic lupus erythematosus. Human Molecular Genetics, 2018, 27, 421-429.	2.9	52
44	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
45	Cardiac Genetic Predisposition in SuddenÂInfant Death Syndrome. Journal of the American College of Cardiology, 2018, 71, 1217-1227.	2.8	66
46	A genome-wide association study for extremely high intelligence. Molecular Psychiatry, 2018, 23, 1226-1232.	7.9	54
47	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
48	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
49	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
50	Exome-Wide Rare Variant Analyses in Sudden Infant Death Syndrome. Journal of Pediatrics, 2018, 203, 423-428.e11.	1.8	17
51	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
52	Mutation in GNE is associated with severe congenital thrombocytopenia. Blood, 2018, 132, 1855-1858.	1.4	46
53	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
54	De novo DNM1L mutation associated with mitochondrial epilepsy syndrome with fever sensitivity. Neurology: Genetics, 2018, 4, e258.	1.9	27

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55	Importance of Variant Interpretation in Whole-Exome Molecular Autopsy. Circulation, 2018, 137, 2705-2715.	1.6	36
56	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
57	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
58	Fine mapping genetic associations between the HLA region and extremely high intelligence. Scientific Reports, 2017, 7, 41182.	3.3	1
59	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. American Journal of Human Genetics, 2017, 100, 364-370.	6.2	32
60	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
61	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
62	Genetic architecture of acne vulgaris. Journal of the European Academy of Dermatology and Venereology, 2017, 31, 1978-1990.	2.4	39
63	MAGI2 Mutations Cause Congenital Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2017, 28, 1614-1621.	6.1	59
64	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
65	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
66	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
67	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
68	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
69	Tissue and Circulating MicroRNA Co-expression Analysis Shows Potential Involvement of miRNAs in the Pathobiology ofÂFrontal Fibrosing Alopecia. Journal of Investigative Dermatology, 2017, 137, 2440-2443.	0.7	8
70	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
71	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
72	Acne and Telomere Length: A New Spectrum between Senescence and Apoptosis Pathways. Journal of Investigative Dermatology, 2017, 137, 513-515.	0.7	6

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73	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
74	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
75	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
76	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
77	Erythrokeratoderma Variabilis Caused by p.Gly45Glu in Connexin 31: Importance of the First Extracellular Loop Glycine Residue for Gap Junction Function. Acta Dermato-Venereologica, 2016, 96, 557-559.	1.3	5
78	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
79	Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. Haematologica, 2016, 101, 1170-1179.	3.5	119
80	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
81	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. Human Mutation, 2016, 37, 1157-1161.	2.5	17
82	Frontal fibrosing alopecia: reflections and hypotheses on aetiology and pathogenesis. Experimental Dermatology, 2016, 25, 847-852.	2.9	66
83	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
84	Ectodermal dysplasiaâ€“skin fragility syndrome resulting from a new atypical homozygous cryptic acceptor splice site mutation in PKP1. Journal of Dermatological Science, 2016, 84, 210-212.	1.9	3
85	Candidate driver genes involved in genome maintenance and DNA repair in SÃ©zary syndrome. Blood, 2016, 127, 3387-3397.	1.4	96
86	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601.	12.8	233
87	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
88	Large Intragenic KRT1 Deletion Underlying Atypical Autosomal Dominant KeratinopathicÂIchthyosis. Journal of Investigative Dermatology, 2016, 136, 2095-2098.	0.7	8
89	Assessment of Minimal Residual Disease in Standard-Risk AML. New England Journal of Medicine, 2016, 374, 422-433.	27.0	662
90	Bi-allelic nonsense mutations inABHD5 underlie a mild phenotype of Dorfman-Chanarin syndrome. Journal of Dermatological Science, 2016, 81, 134-136.	1.9	13

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91	Whole exome sequence analysis reveals a homozygous mutation in PNPLA2 as the cause of severe dilated cardiomyopathy secondary to neutral lipid storage disease. International Journal of Cardiology, 2016, 210, 41-44.	1.7	8
92	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
93	A genome-wide analysis of putative functional and exonic variation associated with extremely high intelligence. Molecular Psychiatry, 2016, 21, 1145-1151.	7.9	20
94	EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088.	8.2	83
95	<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
96	Founder mutation in dystonin-e underlying autosomal recessive epidermolysis bullosa simplex in Kuwait. British Journal of Dermatology, 2015, 172, 527-531.	1.5	29
97	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
98	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
99	Exome Sequencing of 75 Individuals from Multiply Affected Coeliac Families and Large Scale Resequencing Follow Up. PLoS ONE, 2015, 10, e0116845.	2.5	8
100	Germline Mutations in the CDKN2B Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. Cancer Discovery, 2015, 5, 723-729.	9.4	88
101	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
102	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	7.6	81
103	Network-Informed Gene Ranking Tackles Genetic Heterogeneity in Exome-Sequencing Studies of Monogenic Disease. Human Mutation, 2015, 36, 1135-1144.	2.5	7
104	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
105	Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome. American Journal of Human Genetics, 2015, 96, 153-161.	6.2	88
106	Whole-exome sequencing diagnosis of two autosomal recessive disorders in one family. British Journal of Dermatology, 2015, 172, 1407-1411.	1.5	21
107	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
108	Novel indel mutation of STS underlies a new phenotype of self-healing recessive X-linked ichthyosis. Journal of Dermatological Science, 2015, 79, 317-319.	1.9	5

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109	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
110	Novel mutations in PIEZO1 cause an autosomal recessive generalized lymphatic dysplasia with non-immune hydrops fetalis. Nature Communications, 2015, 6, 8085.	12.8	247
111	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
112	Thinking positively: The genetics of high intelligence. Intelligence, 2015, 48, 123-132.	3.0	27
113	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
114	Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. British Journal of Dermatology, 2015, 172, 94-100.	1.5	74
115	SLFN14 mutations underlie thrombocytopenia with excessive bleeding and platelet secretion defects. Journal of Clinical Investigation, 2015, 125, 3600-3605.	8.2	71
116	Whole Exome Sequencing of Flow Purified Tumour Cells Reveals Recurrently Mutated Genes and Pathways in Adult T-Cell Lymphoma/Leukaemia (ATLL). Blood, 2015, 126, 1469-1469.	1.4	1
117	Germline FH Mutations Presenting With Pheochromocytoma. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2046-E2050.	3.6	147
118	Generalized Pustular Eruptions: Time to Adapt the Disease Taxonomy to the Genetic Architecture?. Journal of Investigative Dermatology, 2014, 134, 580-581.	0.7	5
119	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
120	Mutations in GRHL2 Result in an Autosomal-Recessive Ectodermal Dysplasia Syndrome. American Journal of Human Genetics, 2014, 95, 308-314.	6.2	48
121	The ErbB4 CYT2 variant protects EGFR from ligand-induced degradation to enhance cancer cell motility. Science Signaling, 2014, 7, ra78.	3.6	34
122	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, 2014, 23, 2232-2233.	2.9	0
123	Next generation diagnostics of heritable connective tissue disorders. Matrix Biology, 2014, 33, 35-40.	3.6	11
124	Germline CDH1 mutations in bilateral lobular carcinoma in situ. British Journal of Cancer, 2014, 110, 1053-1057.	6.4	70
125	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
126	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308

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127	Epithelial Inflammation Resulting from an Inherited Loss-of-Function Mutation in EGFR. Journal of Investigative Dermatology, 2014, 134, 2570-2578.	0.7	71
128	Genome-wide association study identifies three novel susceptibility loci for severe Acne vulgaris. Nature Communications, 2014, 5, 4020.	12.8	68
129	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
130	Mutations in TJP2 cause progressive cholestatic liver disease. Nature Genetics, 2014, 46, 326-328.	21.4	244
131	Molecular Detection of Minimal Residual Disease Provides the Most Powerful Independent Prognostic Factor Irrespective of Clonal Architecture in Nucleophosmin (NPM1) Mutant Acute Myeloid Leukemia. Blood, 2014, 124, 70-70.	1.4	8
132	Abstract 18543: Whole Exome Sequencing in Sudden Infant Death Syndrome Identifies a High Proportion of Putative Pathogenic and Functionally Significant Rare Variants Related to Inherited Cardiac Conditions. Circulation, 2014, 130, .	1.6	0
133	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
134	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
135	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
136	Impact of next generation sequencing on diagnostics in a genetic skin disease clinic. Experimental Dermatology, 2013, 22, 825-831.	2.9	56
137	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	2.0	85
138	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
139	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
140	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
141	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
142	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
143	Deficiency of terminal ADP-ribose protein glycohydrolase TARG1/C6orf130 in neurodegenerative disease. EMBO Journal, 2013, 32, 1225-1237.	7.8	263
144	Rare Pathogenic Variants in IL36RN Underlie a Spectrum of Psoriasis-Associated Pustular Phenotypes. Journal of Investigative Dermatology, 2013, 133, 1366-1369.	0.7	140

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145	Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. Nature, 2013, 498, 232-235.	27.8	184
146	Î³-Secretase Mutations in Hidradenitis Suppurativa: New Insights into Disease Pathogenesis. Journal of Investigative Dermatology, 2013, 133, 601-607.	0.7	133
147	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
148	The future of genomics for developmentalists. Development and Psychopathology, 2013, 25, 1263-1278.	2.3	41
149	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. Nature Genetics, 2013, 45, 1244-1248.	21.4	289
150	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
151	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	21.4	231
152	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
153	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
154	A NovelABCA12Mutation in Two Families with Congenital Ichthyosis. Scientifica, 2012, 2012, 1-6.	1.7	6
155	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
156	Next-Generation Diagnostics for Genodermatoses. Journal of Investigative Dermatology, 2012, 132, E27-E28.	0.7	8
157	De Novo Mutations in MLL Cause Wiedemann-Steiner Syndrome. American Journal of Human Genetics, 2012, 91, 358-364.	6.2	225
158	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
159	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
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
161	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
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