Michael A Simpson

List of Publications by Year in Descending Order

186 papers	9,803 citations	55 h-index	94 g-index
196 ext. papers	11,708 ext. citations	8 avg, IF	5.42 L-index

#	Paper	IF	Citations
186	Genome-wide association meta-analysis identifies 29 new acne susceptibility loci.. *Nature Communications*, **2022**, 13, 702	17.4	1
185	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	6	1
184	DIFFERENCES IN CLINICAL FEATURES AND COMORBID BURDEN BETWEEN HLA-C*06:02 CARRIER GROUPS IN MORE THAN 9,000 PEOPLE WITH PSORIASIS. *Journal of Investigative Dermatology*, **2021** ,	4.3	0
183	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	3.7	
182	Application of information theoretic feature selection and machine learning methods for the development of genetic risk prediction models. *Scientific Reports*, **2021**, 11, 23335	4.9	2
181	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	4.9	6
180	Autosomal recessive hypotrichosis with loose anagen hairs associated with TKFC mutations. *British Journal of Dermatology*, **2021**, 184, 935-943	4	2
179	A germline mutation in the platelet-derived growth factor receptor beta gene may be implicated in hereditary progressive mucinous histiocytosis. *British Journal of Dermatology*, **2021**, 184, 967-970	4	5
178	Patients with triple-negative, JAK2V617F- and CALR-mutated essential thrombocythemia share a unique gene expression signature. *Blood Advances*, **2021**, 5, 1059-1068	7.8	3
177	New Homozygous Missense MSMO1 Mutation in Two Siblings with SC4MOL Deficiency Presenting with Psoriasiform Dermatitis. *Cytogenetic and Genome Research*, **2020**, 160, 523-530	1.9	1
176	Phenotypic and genetic spectrum of epilepsy with myoclonic atonic seizures. *Epilepsia*, **2020**, 61, 995-1007	6.4	18
175	Molecular basis and inheritance patterns of amyloidosis cutis dyschromica. *Clinical and Experimental Dermatology*, **2020**, 45, 650-653	1.8	5
174	Mutations in SPATA13/ASEF2 cause primary angle closure glaucoma. *PLoS Genetics*, **2020**, 16, e1008721	6	7
173	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	11.5	29
172	Psoriasis and Genetics. *Acta Dermato-Venereologica*, **2020**, 100, adv00030	2.2	20
171	Coagulation Factor XIII-A Subunit Missense Mutation in the Pathobiology of Autosomal Dominant Multiple Dermatofibromas. *Journal of Investigative Dermatology*, **2020**, 140, 624-635.e7	4.3	4
170	Homozygous Nonsense Mutation in DSC3 Resulting in Skin Fragility and Hypotrichosis. *Journal of Investigative Dermatology*, **2020**, 140, 1285-1288	4.3	4

169	Blaschko-linear lichen planus: Clinicopathological and genetic analysis. *Journal of Dermatology*, **2020**, 47, e384-e385	1.6	0
168	Genotype-phenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. *British Journal of Dermatology*, **2020**, 182, 729-737	4	19
167	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. *Clinical and Experimental Dermatology*, **2020**, 45, 391-394	1.8	1
166	The ADAMTS13-VWF axis is dysregulated in chronic thromboembolic pulmonary hypertension. *European Respiratory Journal*, **2019**, 53,	13.6	16
165	Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset. *Npj Parkinsonls Disease*, **2019**, 5, 8	9.7	47
164	Frequency of pathogenic germline variants in BRCA1, BRCA2, PALB2, CHEK2 and TP53 in ductal carcinoma in situ diagnosed in women under the age of 50 years. *Breast Cancer Research*, **2019**, 21, 58	8.3	7
163	Sequencing of human genomes with nanopore technology. *Nature Communications*, **2019**, 10, 1869	17.4	89
162	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	5.3	24
161	Genome-wide association study in frontal fibrosing alopecia identifies four susceptibility loci including HLA-B*07:02. *Nature Communications*, **2019**, 10, 1150	17.4	55
160	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17
159	Novel ADA2 mutation presenting with neutropenia, lymphopenia and bone marrow failure in patients with deficiency in adenosine deaminase 2 (DADA2). *British Journal of Haematology*, **2019**, 186, e60-e64	4.5	8
158	Noncardiac genetic predisposition in sudden infant death syndrome. *Genetics in Medicine*, **2019**, 21, 641-649	6.4	6
157	Frequency of Pathogenic Germline Variants in , and in Sporadic Lobular Breast Cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2019**, 28, 1162-1168	4	12
156	Semidominant GPNMB Mutations in Amyloidosis Cutis Dyschromica. *Journal of Investigative Dermatology*, **2019**, 139, 2550-2554.e9	4.3	7
155	Molecular, Epigenetic and Gene Expression Profiling of Triple Negative Essential Thrombocythaemia. *Blood*, **2019**, 134, 308-308	2.2	
154	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	11.5	63
153	Consanguinity and Double Recessive Gene Pathology: Cutis Laxa (PYCR1) and Nephrotic Syndrome (PLCE1). *JAMA Dermatology*, **2019**, 155, 257-259	5.1	4
152	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	4.7	3

151	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. *American Journal of Human Genetics*, **2018**, 102, 494-504	11	44
150	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	40	49
149	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	4.3	7
148	Recessive Mutation in FAM83G Associated with Palmoplantar Keratoderma and Exuberant Scalp Hair. *Journal of Investigative Dermatology*, **2018**, 138, 984-987	4.3	9
147	MED12, TERT promoter and RBM15 mutations in primary and recurrent phyllodes tumours. *British Journal of Cancer*, **2018**, 118, 277-284	8.7	22
146	De novo mutations implicate novel genes in systemic lupus erythematosus. *Human Molecular Genetics*, **2018**, 27, 421-429	5.6	29
145	Exome Sequencing and Genotyping Identify a Rare Variant in NLRP7 Gene Associated With Ulcerative Colitis. *Journal of Crohnls and Colitis*, **2018**, 12, 321-326	1.5	7
144	Cardiac Genetic Predisposition in Sudden Infant Death Syndrome. *Journal of the American College of Cardiology*, **2018**, 71, 1217-1227	15.1	43
143	A genome-wide association study for extremely high intelligence. *Molecular Psychiatry*, **2018**, 23, 1226-1232	15.1	35
142	Exome Sequencing and Rare Variant Analysis Reveals Multiple Filaggrin Mutations in Bangladeshi Families with Atopic Eczema and Additional Risk Genes. *Journal of Investigative Dermatology*, **2018**, 138, 2674-2677	4.3	19
141	De novo mutation associated with mitochondrial epilepsy syndrome with fever sensitivity. *Neurology: Genetics*, **2018**, 4, e258	3.8	18
140	Importance of Variant Interpretation in Whole-Exome Molecular Autopsy: Population-Based Case Series. *Circulation*, **2018**, 137, 2705-2715	16.7	23
139	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. *JAMA Neurology*, **2018**, 75, 105-113	17.2	20
138	Epistemic uncertainties and natural hazard risk assessment Part 2: What should constitute good practice?. *Natural Hazards and Earth System Sciences*, **2018**, 18, 2769-2783	3.9	22
137	Genome-wide meta-analysis implicates mediators of hair follicle development and morphogenesis in risk for severe acne. *Nature Communications*, **2018**, 9, 5075	17.4	18
136	Exome-Wide Rare Variant Analyses in Sudden Infant Death Syndrome. *Journal of Pediatrics*, **2018**, 203, 423-428.e11	3.6	12
135	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
134	Mutation in is associated with severe congenital thrombocytopenia. *Blood*, **2018**, 132, 1855-1858	2.2	30

133	Genomic and clinical profiling of a national nephrotic syndrome cohort advocates a precision medicine approach to disease management. *Kidney International*, **2017**, 91, 937-947	9.9	130
132	Congenital Anonychia and Uncombable Hair Syndrome: Coinheritance of Homozygous Mutations in RSPO4 and PADI3. *Journal of Investigative Dermatology*, **2017**, 137, 1176-1179	4.3	13
131	Fine mapping genetic associations between the HLA region and extremely high intelligence. *Scientific Reports*, **2017**, 7, 41182	4.9	1
130	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. *American Journal of Human Genetics*, **2017**, 100, 364-370	11	22
129	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. *Nature Genetics*, **2017**, 49, 537-549	36.3	52
128	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. *Science Translational Medicine*, **2017**, 9,	17.5	74
127	Genetic architecture of acne vulgaris. *Journal of the European Academy of Dermatology and Venereology*, **2017**, 31, 1978-1990	4.6	27
126	Mutations Cause Congenital Nephrotic Syndrome. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 1614-1621	12.7	49
125	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	2.8	7
124	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. *Neurogenetics*, **2017**, 18, 227-235	3	5
123	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	2.4	36
122	Novel homozygous missense mutation in NT5C2 underlying hereditary spastic paraplegia SPG45. *American Journal of Medical Genetics, Part A*, **2017**, 173, 3109-3113	2.5	11
121	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in a Spectrum of Keratinization Disorders Associated with Thrombocytopenia. *Journal of Investigative Dermatology*, **2017**, 137, 2344-2353	4.3	35
120	Tissue and Circulating MicroRNA Co-expression Analysis Shows Potential Involvement of miRNAs in the Pathobiology of Frontal Fibrosing Alopecia. *Journal of Investigative Dermatology*, **2017**, 137, 2440-2443	4.3	7
119	PIK3CA mutations are common in lobular carcinoma in situ, but are not a biomarker of progression. *Breast Cancer Research*, **2017**, 19, 7	8.3	13
118	Syndromic inherited poikiloderma due to a de novo mutation in FAM111B. *British Journal of Dermatology*, **2017**, 176, 534-536	4	9
117	Acne and Telomere Length: A New Spectrum between Senescence and Apoptosis Pathways. *Journal of Investigative Dermatology*, **2017**, 137, 513-515	4.3	4
116	Pathogenic variants in HTRA2 cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria. *Journal of Inherited Metabolic Disease*, **2017**, 40, 121-130	5.4	16

115	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. *Brain*, **2017**, 140, 940-952	11.2	42
114	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	5.6	25
113	Phenome-wide association study using research participantsR self-reported data provides insight into the Th17 and IL-17 pathway. *PLoS ONE*, **2017**, 12, e0186405	3.7	14
112	Ectodermal dysplasia-skin fragility syndrome resulting from a new atypical homozygous cryptic acceptor splice site mutation in PKP1. *Journal of Dermatological Science*, **2016**, 84, 210-212	4.3	2
111	Candidate driver genes involved in genome maintenance and DNA repair in Sézary syndrome. *Blood* , **2016**, 127, 3387-97	2.2	71
110	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism-dystonia. *Nature Communications*, **2016**, 7, 11601	17.4	160
109	Hidradenitis suppurativa: haploinsufficiency of gamma-secretase components does not affect gamma-secretase enzyme activity in vitro. *British Journal of Dermatology*, **2016**, 175, 632-5	4	8
108	Large Intragenic KRT1 Deletion Underlying Atypical Autosomal Dominant Keratinopathic Ichthyosis. *Journal of Investigative Dermatology*, **2016**, 136, 2095-2098	4.3	3
107	Assessment of Minimal Residual Disease in Standard-Risk AML. *New England Journal of Medicine*, **2016**, 374, 422-33	59.2	500
106	Bi-allelic nonsense mutations inABHD5 underlie a mild phenotype of Dorfman-Chanarin syndrome. *Journal of Dermatological Science*, **2016**, 81, 134-6	4.3	12
105	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, 41-4	3.2	6
104	Germline ESR2 mutation predisposes to medullary thyroid carcinoma and causes up-regulation of RET expression. *Human Molecular Genetics*, **2016**, 25, 1836-45	5.6	21
103	A genome-wide analysis of putative functional and exonic variation associated with extremely high intelligence. *Molecular Psychiatry*, **2016**, 21, 1145-51	15.1	16
102	EPHB4 kinase-inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. *Journal of Clinical Investigation*, **2016**, 126, 3080-8	15.9	58
101	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, 557-9	2.2	3
100	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	11	68
99	Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. *Haematologica*, **2016**, 101, 1170-1179	6.6	89
98	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	2.5	7

97	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. *Human Mutation*, **2016**, 37, 1157-1161	4.7	14
96	Frontal fibrosing alopecia: reflections and hypotheses on aetiology and pathogenesis. *Experimental Dermatology*, **2016**, 25, 847-852	4	57
95	Induced Pluripotent Stem Cell Differentiation and Three-Dimensional Tissue Formation Attenuate Clonal Epigenetic Differences in Trichohyalin. *Stem Cells and Development*, **2016**, 25, 1366-75	4.4	8
94	Whole-exome sequencing diagnosis of two autosomal recessive disorders in one family. *British Journal of Dermatology*, **2015**, 172, 1407-11	4	16
93	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	6	43
92	Novel indel mutation of STS underlies a new phenotype of self-healing recessive X-linked ichthyosis. *Journal of Dermatological Science*, **2015**, 79, 317-9	4.3	5
91	Progressive hyperpigmentation in a Taiwanese child due to an inborn error of vitamin B12 metabolism (cblJ). *British Journal of Dermatology*, **2015**, 172, 1111-5	4	17
90	Novel mutations in PIEZO1 cause an autosomal recessive generalized lymphatic dysplasia with non-immune hydrops fetalis. *Nature Communications*, **2015**, 6, 8085	17.4	174
89	Truncating mutation in intracellular phospholipase A gene (DDHD2) in hereditary spastic paraplegia with intellectual disability (SPG54). *BMC Research Notes*, **2015**, 8, 271	2.3	9
88	Thinking positively: The genetics of high intelligence. *Intelligence*, **2015**, 48, 123-132	3	19
87	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzen-Goldberg syndrome. *European Journal of Human Genetics*, **2015**, 23, 224-8	5.3	42
86	Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. *British Journal of Dermatology*, **2015**, 172, 94-100	4	54
85	EGFR mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. *Molecular Genetics & Genomic Medicine*, **2015**, 3, 452-8	2.3	8
84	Founder mutation in dystonin-e underlying autosomal recessive epidermolysis bullosa simplex in Kuwait. *British Journal of Dermatology*, **2015**, 172, 527-31	4	20
83	The role of SLC2A1 mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of GLUT1 deficiency syndrome. *Epilepsia*, **2015**, 56, e203-8	6.4	52
82	Diversity and impact of rare variants in genes encoding the platelet G protein-coupled receptors. *Thrombosis and Haemostasis*, **2015**, 113, 826-37	7	15
81	Exome sequencing of 75 individuals from multiply affected coeliac families and large scale resequencing follow up. *PLoS ONE*, **2015**, 10, e0116845	3.7	8
80	Germline Mutations in the CDKN2B Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. *Cancer Discovery*, **2015**, 5, 723-9	24.4	61

79	Homozygous missense mutation in IL36RN in generalized pustular dermatosis with intraoral involvement compatible with both AGEP and generalized pustular psoriasis. *JAMA Dermatology*, **2015**, 151, 452-3	5.1	31
78	LRPPRC mutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. *Brain*, **2015**, 138, 3503-19	11.2	63
77	Network-Informed Gene Ranking Tackles Genetic Heterogeneity in Exome-Sequencing Studies of Monogenic Disease. *Human Mutation*, **2015**, 36, 1135-44	4.7	7
76	Use of next-generation sequencing and candidate gene analysis to identify underlying defects in patients with inherited platelet function disorders. *Journal of Thrombosis and Haemostasis*, **2015**, 13, 643-50	15.4	57
75	Defects of CRB2 cause steroid-resistant nephrotic syndrome. *American Journal of Human Genetics*, **2015**, 96, 153-61	11	76
74	SLFN14 mutations underlie thrombocytopenia with excessive bleeding and platelet secretion defects. *Journal of Clinical Investigation*, **2015**, 125, 3600-5	15.9	51
73	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	2.2	1
72	Genetic variation in schlafen genes in a patient with a recapitulation of the murine Elektra phenotype. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1462-5, 1465.e1-5	11.5	4
71	Exome-wide rare variant analysis identifies TUBA4A mutations associated with familial ALS. *Neuron*, **2014**, 84, 324-31	13.9	229
70	Epithelial inflammation resulting from an inherited loss-of-function mutation in EGFR. *Journal of Investigative Dermatology*, **2014**, 134, 2570-2578	4.3	51
69	Genome-wide association study identifies three novel susceptibility loci for severe Acne vulgaris. *Nature Communications*, **2014**, 5, 4020	17.4	48
68	AP1S3 mutations are associated with pustular psoriasis and impaired Toll-like receptor 3 trafficking. *American Journal of Human Genetics*, **2014**, 94, 790-7	11	105
67	Mutations in TJP2 cause progressive cholestatic liver disease. *Nature Genetics*, **2014**, 46, 326-8	36.3	202
66	Germline FH mutations presenting with pheochromocytoma. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E2046-50	5.6	127
65	Generalized pustular eruptions: time to adapt the disease taxonomy to the genetic architecture?. *Journal of Investigative Dermatology*, **2014**, 134, 580-581	4.3	3
64	The missense mutation p.R1303Q in type XVII collagen underlies junctional epidermolysis bullosa resembling Kindler syndrome. *Journal of Investigative Dermatology*, **2014**, 134, 845-849	4.3	16
63	Mutations in GRHL2 result in an autosomal-recessive ectodermal Dysplasia syndrome. *American Journal of Human Genetics*, **2014**, 95, 308-14	11	37
62	The ErbB4 CYT2 variant protects EGFR from ligand-induced degradation to enhance cancer cell motility. *Science Signaling*, **2014**, 7, ra78	8.8	28

61	Next generation diagnostics of heritable connective tissue disorders. *Matrix Biology*, **2014**, 33, 35-40	11.4	9
60	Germline CDH1 mutations in bilateral lobular carcinoma in situ. *British Journal of Cancer*, **2014**, 110, 1053-7	4.7	54
59	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	2.2	3
58	A combination of mutations in AKR1D1 and SKIV2L in a family with severe infantile liver disease. *Orphanet Journal of Rare Diseases*, **2013**, 8, 74	4.2	13
57	Acrocallosal syndrome: identification of a novel KIF7 mutation and evidence for oligogenic inheritance. *European Journal of Medical Genetics*, **2013**, 56, 39-42	2.6	18
56	Acral peeling skin syndrome resulting from a homozygous nonsense mutation in the CSTA gene encoding cystatin A. *Pediatric Dermatology*, **2013**, 30, e87-8	1.9	30
55	Impact of next generation sequencing on diagnostics in a genetic skin disease clinic. *Experimental Dermatology*, **2013**, 22, 825-31	4	41
54	MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. *Clinical Genetics*, **2013**, 84, 539-45	4	66
53	Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. *Nature Genetics*, **2013**, 45, 1300-8	36.3	172
52	Enrichment of FLI1 and RUNX1 mutations in families with excessive bleeding and platelet dense granule secretion defects. *Blood*, **2013**, 122, 4090-3	2.2	90
51	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, 1395-403	5.6	87
50	Predicting the functional consequences of non-synonymous DNA sequence variants--evaluation of bioinformatics tools and development of a consensus strategy. *Genomics*, **2013**, 102, 223-8	4.3	76
49	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, 346-56	11	126
48	Deficiency of terminal ADP-ribose protein glycohydrolase TARG1/C6orf130 in neurodegenerative disease. *EMBO Journal*, **2013**, 32, 1225-37	13	215
47	Rare pathogenic variants in IL36RN underlie a spectrum of psoriasis-associated pustular phenotypes. *Journal of Investigative Dermatology*, **2013**, 133, 1366-9	4.3	109
46	Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. *Nature*, **2013**, 498, 232-5	50.4	156
45	Secretase mutations in hidradenitis suppurativa: new insights into disease pathogenesis. *Journal of Investigative Dermatology*, **2013**, 133, 601-607	4.3	104
44	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, 956-60	15.7	120

43	The future of genomics for developmentalists. *Development and Psychopathology*, **2013**, 25, 1263-78	4.3	34
42	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. *Nature Genetics*, **2013**, 45, 1244-1248	36.3	217
41	Next generation exome sequencing of paediatric inflammatory bowel disease patients identifies rare and novel variants in candidate genes. *Gut*, **2013**, 62, 977-84	19.2	92
40	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. *Nature Genetics*, **2013**, 45, 83-7	36.3	192
39	Rare variations in IL36RN in severe adverse drug reactions manifesting as acute generalized exanthematous pustulosis. *Journal of Investigative Dermatology*, **2013**, 133, 1904-7	4.3	77
38	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	3.7	33
37	Recurrent heterozygous missense mutation, p.Gly573Ser, in the TRPV3 gene in an Indian boy with sporadic Olmsted syndrome. *British Journal of Dermatology*, **2012**, 167, 440-2	4	55
36	De novo mutations in MLL cause Wiedemann-Steiner syndrome. *American Journal of Human Genetics*, **2012**, 91, 358-64	11	166
35	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. *American Journal of Human Genetics*, **2012**, 91, 1115-21	11	48
34	Mutations in the □ecretase genes NCSTN, PSENEN, and PSEN1 underlie rare forms of hidradenitis suppurativa (acne inversa). *Journal of Investigative Dermatology*, **2012**, 132, 2459-2461	4.3	91
33	Identification of rare, disease-associated variants in the promoter region of the RNF114 psoriasis susceptibility gene. *Journal of Investigative Dermatology*, **2012**, 132, 1297-9	4.3	4
32	De novo mutations of the gene encoding the histone acetyltransferase KAT6B cause Genitopatellar syndrome. *American Journal of Human Genetics*, **2012**, 90, 290-4	11	77
31	Mutations in KIF11 cause autosomal-dominant microcephaly variably associated with congenital lymphedema and chorioretinopathy. *American Journal of Human Genetics*, **2012**, 90, 356-62	11	108
30	A Novel ABCA12 Mutation in Two Families with Congenital Ichthyosis. *Scientifica*, **2012**, 2012, 649090	2.6	6
29	Serpentine fibula polycystic kidney syndrome is part of the phenotypic spectrum of Hajdu-Cheney syndrome. *European Journal of Human Genetics*, **2012**, 20, 122-4	5.3	50
28	Next-generation diagnostics for genodermatoses. *Journal of Investigative Dermatology*, **2012**, 132, E27-8	4.3	5
27	Massively parallel sequencing and identification of genes for primary lymphoedema: a perfect fit. *Clinical Genetics*, **2011**, 80, 110-6	4	4
26	Mutations in NOTCH2 cause Hajdu-Cheney syndrome, a disorder of severe and progressive bone loss. *Nature Genetics*, **2011**, 43, 303-5	36.3	242

25	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, 432-7	11	362
24	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, 574-85	11	87
23	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, 251-5	5.8	77
22	PSENEN and NCSTN mutations in familial hidradenitis suppurativa (Acne Inversa). *Journal of Investigative Dermatology*, **2011**, 131, 1568-70	4.3	82
21	Mutations in GATA2 cause primary lymphedema associated with a predisposition to acute myeloid leukemia (Emberger syndrome). *Nature Genetics*, **2011**, 43, 929-31	36.3	358
20	Raine syndrome: a clinical, radiographic and genetic investigation of a case from the Indian subcontinent. *Clinical Dysmorphology*, **2010**, 19, 153-156	0.9	16
19	Defective mitochondrial mRNA maturation is associated with spastic ataxia. *American Journal of Human Genetics*, **2010**, 87, 655-60	11	72
18	A three-stage genome-wide association study of general cognitive ability: hunting the small effects. *Behavior Genetics*, **2010**, 40, 759-67	3.2	61
17	Lethal cystic kidney disease in Amish neonates associated with homozygous nonsense mutation of NPHP3. *American Journal of Kidney Diseases*, **2009**, 53, 790-5	7.4	11
16	A mutation in NFkappaB interacting protein 1 causes cardiomyopathy and woolly haircoat syndrome of Poll Hereford cattle. *Animal Genetics*, **2009**, 40, 42-6	2.5	25
15	Mutations in FAM20C also identified in non-lethal osteosclerotic bone dysplasia. *Clinical Genetics*, **2009**, 75, 271-6	4	81
14	Homozygous mutation of desmocollin-2 in arrhythmogenic right ventricular cardiomyopathy with mild palmoplantar keratoderma and woolly hair. *Cardiology*, **2009**, 113, 28-34	1.6	73
13	A novel locus for an autosomal recessive hereditary spastic paraplegia (SPG35) maps to 16q21-q23. *Neurology*, **2008**, 71, 248-52	6.5	39
12	Sequence alterations within CYP7B1 implicate defective cholesterol homeostasis in motor-neuron degeneration. *American Journal of Human Genetics*, **2008**, 82, 510-5	11	148
11	Homozygous mutation of MYBPC3 associated with severe infantile hypertrophic cardiomyopathy at high frequency among the Amish. *Heart*, **2008**, 94, 1326-30	5.1	48
10	Novel mutations in the pejvakin gene are associated with autosomal recessive non-syndromic hearing loss in Iranian families. *Clinical Genetics*, **2007**, 72, 261-3	4	26
9	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, 906-12	11	149
8	Endogenous spartin, mutated in hereditary spastic paraplegia, has a complex subcellular localization suggesting diverse roles in neurons. *Experimental Cell Research*, **2006**, 312, 2764-77	4.2	27

7	A novel NIPA1 mutation associated with a pure form of autosomal dominant hereditary spastic paraplegia. *Neurogenetics*, **2005**, 6, 79-84	3	46
6	Novel mutation in desmoplakin causes arrhythmogenic left ventricular cardiomyopathy. *Circulation*, **2005**, 112, 636-42	16.7	217
5	A new locus for autosomal recessive complicated hereditary spastic paraplegia (SPG26) maps to chromosome 12p11.1-12q14. *Journal of Medical Genetics*, **2005**, 42, 80-2	5.8	33
4	Complicated hereditary spastic paraplegia with thin corpus callosum (HSP-TCC) and childhood onset. *Neuropediatrics*, **2005**, 36, 274-8	1.6	20
3	Infantile-onset symptomatic epilepsy syndrome caused by a homozygous loss-of-function mutation of GM3 synthase. *Nature Genetics*, **2004**, 36, 1225-9	36.3	304
2	Maspardin is mutated in mast syndrome, a complicated form of hereditary spastic paraplegia associated with dementia. *American Journal of Human Genetics*, **2003**, 73, 1147-56	11	145
1	How to Design and Use a Questionnaire in Evaluation and Educational Research. *Medical Teacher*, **1984**, 6, 122-7	3	2