W H Irwin Mclean

List of Publications by Year
in descending order

251 papers	28,889 citations	4942 84 h-index	5519 163 g-index
257 all docs	257 docs citations	257 times ranked	18060 citing authors

#	Article	IF	Citations
1	Common loss-of-function variants of the epidermal barrier protein filaggrin are a major predisposing factor for atopic dermatitis. Nature Genetics, 2006, 38, 441-446.	9.4	2,584
2	Filaggrin Mutations Associated with Skin and Allergic Diseases. New England Journal of Medicine, 2011, 365, 1315-1327.	13.9	996
3	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	9.4	918
4	Loss-of-function mutations in the gene encoding filaggrin cause ichthyosis vulgaris. Nature Genetics, 2006, 38, 337-342.	9.4	916
5	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
6	Filaggrin in the frontline: role in skin barrier function and disease. Journal of Cell Science, 2009, 122, 1285-1294.	1.2	672
7	Comprehensive analysis of the gene encoding filaggrin uncovers prevalent and rare mutations in ichthyosis vulgaris and atopic eczema. Nature Genetics, 2007, 39, 650-654.	9.4	598
8	Emollient enhancement of the skin barrier from birth offers effective atopic dermatitis prevention. Journal of Allergy and Clinical Immunology, 2014, 134, 818-823.	1.5	594
9	Loss-of-function variations within the filaggrin gene predispose for atopic dermatitis with allergic sensitizations. Journal of Allergy and Clinical Immunology, 2006, 118, 214-219.	1.5	567
10	A homozygous frameshift mutation in the mouse Flg gene facilitates enhanced percutaneous allergen priming. Nature Genetics, 2009, 41, 602-608.	9.4	438
11	Intermediate Filament Proteins and Their Associated Diseases. New England Journal of Medicine, 2004, 351, 2087-2100.	13.9	434
12	One Remarkable Molecule: Filaggrin. Journal of Investigative Dermatology, 2012, 132, 751-762.	0.3	433
13	Loss-of-function variants in the filaggrin gene are a significant risk factor for peanut allergy. Journal of Allergy and Clinical Immunology, 2011, 127, 661-667.	1.5	424
14	Human keratin diseases: the increasing spectrum of disease and subtlety of the phenotype-genotype correlation. British Journal of Dermatology, 1999, 140, 815-828.	1.4	413
15	Filaggrin in atopic dermatitis. Journal of Allergy and Clinical Immunology, 2008, 122, 689-693.	1.5	410
16	Plectin deficiency results in muscular dystrophy with epidermolysis bullosa. Nature Genetics, 1996, 13, 450-457.	9.4	394
17	Filaggrin mutations, atopic eczema, hay fever, and asthma in children. Journal of Allergy and Clinical Immunology, 2008, 121, 1203-1209.e1.	1.5	380
18	The burden of disease associated with filaggrin mutations: A population-based, longitudinal birth cohort study. Journal of Allergy and Clinical Immunology, 2008, 121, 872-877.e9.	1.5	318

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19	The Human Intermediate Filament Database: comprehensive information on a gene family involved in many human diseases. Human Mutation, 2008, 29, 351-360.	1.1	309
20	Loss of Kindlin-1, a Human Homolog of the Caenorhabditis elegans Actinâ€“Extracellular-Matrix Linker Protein UNC-112, Causes Kindler Syndrome. American Journal of Human Genetics, 2003, 73, 174-187.	2.6	305
21	Null Mutations in the Filaggrin Gene (FLG) Determine Major Susceptibility to Early-Onset Atopic Dermatitis that Persists into Adulthood. Journal of Investigative Dermatology, 2007, 127, 564-567.	0.3	298
22	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. Nature Genetics, 2013, 45, 1244-1248.	9.4	289
23	Atopic dermatitis increases the effect of exposure to peanut antigen in dust on peanut sensitization and likely peanut allergy. Journal of Allergy and Clinical Immunology, 2015, 135, 164-170.e4.	1.5	280
24	Skin microbiome before development of atopic dermatitis: Early colonization with commensal staphylococci at 2Âmonths is associated with a lower risk of atopic dermatitis at 1Âyear. Journal of Allergy and Clinical Immunology, 2017, 139, 166-172.	1.5	276
25	Loss-of-Function Mutations in the Filaggrin Gene Lead to Reduced Level of Natural Moisturizing Factor in the Stratum Corneum. Journal of Investigative Dermatology, 2008, 128, 2117-2119.	0.3	273
26	Levels of filaggrin degradation products are influenced by both filaggrin genotype and atopic dermatitis severity. Allergy: European Journal of Allergy and Clinical Immunology, 2011, 66, 934-940.	2.7	251
27	Lipoid proteinosis maps to 1q21 and is caused by mutations in the extracellular matrix protein 1 gene (ECM1). Human Molecular Genetics, 2002, 11, 833-840.	1.4	246
28	Filaggrin in atopic dermatitis. Journal of Allergy and Clinical Immunology, 2009, 124, R2-R6.	1.5	245
29	Peanut allergy: Effect of environmental peanut exposure in children with filaggrin loss-of-function mutations. Journal of Allergy and Clinical Immunology, 2014, 134, 867-875.e1.	1.5	240
30	Unique mutations in the filaggrin gene in Japanese patients with ichthyosis vulgaris andÂatopic dermatitis. Journal of Allergy and Clinical Immunology, 2007, 119, 434-440.	1.5	233
31	Filaggrin loss-of-function mutations are associated with enhanced expression of IL-1 cytokines in the stratum corneum of patients with atopic dermatitis and in a murine model of filaggrin deficiency. Journal of Allergy and Clinical Immunology, 2012, 129, 1031-1039.e1.	1.5	226
32	Gene-Environment Interaction in the Onset of Eczema in Infancy: Filaggrin Loss-of-Function Mutations Enhanced by Neonatal Cat Exposure. PLoS Medicine, 2008, 5, e131.	3.9	215
33	Filaggrin Genotype in Ichthyosis Vulgaris Predicts Abnormalities in Epidermal Structure and Function. American Journal of Pathology, 2011, 178, 2252-2263.	1.9	213
34	Breaking the (Un)Sound Barrier: Filaggrin Is a Major Gene for Atopic Dermatitis. Journal of Investigative Dermatology, 2006, 126, 1200-1202.	0.3	212
35	Prevalent and Rare Mutations in the Gene Encoding Filaggrin Cause Ichthyosis Vulgaris and Predispose Individuals to Atopic Dermatitis. Journal of Investigative Dermatology, 2006, 126, 1770-1775.	0.3	210
36	Mutations in cornea-specific keratin K3 or K12 genes cause Meesmann's corneal dystrophy. Nature Genetics, 1997, 16, 184-187.	9.4	206

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37	Filaggrin loss-of-function mutations are associated with early-onset eczema, eczema severity and transepidermal water loss at 3â€ƒmonths of age. British Journal of Dermatology, 2010, 163, 1333-1336.	1.4	206
38	Human keratin diseases:. Hereditary fragility of specific epithelial tissues. Experimental Dermatology, 1996, 5, 297-307.	1.4	205
39	A genome-wide association study of atopic dermatitis identifies loci with overlapping effects on asthma and psoriasis. Human Molecular Genetics, 2013, 22, 4841-4856.	1.4	202
40	Filaggrin null mutations are associated with increased asthma severity in children and young adults. Journal of Allergy and Clinical Immunology, 2007, 120, 64-68.	1.5	199
41	The persistence of atopic dermatitis and filaggrin (FLG) mutations in a US longitudinal cohort. Journal of Allergy and Clinical Immunology, 2012, 130, 912-917.	1.5	193
42	Loss-of-function mutations of an inhibitory upstream ORF in the human hairless transcript cause Marie Unna hereditary hypotrichosis. Nature Genetics, 2009, 41, 228-233.	9.4	190
43	Intragenic Copy Number Variation within Filaggrin Contributes to the Risk of Atopic Dermatitis with a Dose-Dependent Effect. Journal of Investigative Dermatology, 2012, 132, 98-104.	0.3	185
44	Clinical and Pathological Features of Pachyonychia Congenita. Journal of Investigative Dermatology Symposium Proceedings, 2005, 10, 3-17.	0.8	176
45	High-density genotyping study identifies four new susceptibility loci for atopic dermatitis. Nature Genetics, 2013, 45, 808-812.	9.4	167
46	Atopic dermatitis is associated with an increased risk for rheumatoid arthritis and inflammatory bowel disease, and a decreased risk for type 1 diabetes. Journal of Allergy and Clinical Immunology, 2016, 137, 130-136.	1.5	166
47	Genome-wide Comparative Analysis of Atopic Dermatitis and Psoriasis Gives Insight into Opposing Genetic Mechanisms. American Journal of Human Genetics, 2015, 96, 104-120.	2.6	163
48	A Mutation in Human Keratin K6b Produces a Phenocopy of the K17 Disorder Pachyonychia Congenita Type 2. Human Molecular Genetics, 1998, 7, 1143-1148.	1.4	161
49	Atopic Dermatitis and Disease Severity Are the Main Risk Factors for Food Sensitization in Exclusively Breastfed Infants. Journal of Investigative Dermatology, 2014, 134, 345-350.	0.3	158
50	Clinical Severity Correlates with Impaired Barrier in Filaggrin-Related Eczema. Journal of Investigative Dermatology, 2009, 129, 682-689.	0.3	154
51	Filaggrin null mutations and childhood atopic eczema: A population-based case-control study. Journal of Allergy and Clinical Immunology, 2008, 121, 940-946.e3.	1.5	143
52	Raman profiles of the stratum corneum define 3 filaggrin genotypeâ€“determined atopic dermatitis endophenotypes. Journal of Allergy and Clinical Immunology, 2010, 126, 574-580.e1.	1.5	140
53	Eczema Genetics: Current State of Knowledge and Future Goals. Journal of Investigative Dermatology, 2009, 129, 543-552.	0.3	139
54	Homozygous deletion mutations in the plectin gene (PLEC1) in patients with epidermolysis bullosa simplex associated with late-onset muscular dystrophy. Human Molecular Genetics, 1996, 5, 1539-1546.	1.4	136

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55	Tmem79/Matt is the matted mouse gene and is a predisposing gene for atopic dermatitis in human subjects. Journal of Allergy and Clinical Immunology, 2013, 132, 1121-1129.	1.5	135
56	Compound Heterozygosity for Non-Sense and Mis-Sense Mutations in Desmoplakin Underlies Skin Fragility/Woolly Hair Syndrome. Journal of Investigative Dermatology, 2002, 118, 232-238.	0.3	129
57	Specific Filaggrin Mutations Cause Ichthyosis Vulgaris and Are Significantly Associated with Atopic Dermatitis in Japan. Journal of Investigative Dermatology, 2008, 128, 1436-1441.	0.3	128
58	Filaggrin mutations in the onset of eczema, sensitization, asthma, hay fever and the interaction with cat exposure. Allergy: European Journal of Allergy and Clinical Immunology, 2009, 64, 1758-1765.	2.7	127
59	Impact of atopic dermatitis and loss-of-function mutations in the filaggrin gene on the development of occupational irritant contact dermatitis. British Journal of Dermatology, 2013, 168, 326-332.	1.4	125
60	An unusual N-terminal deletion of the laminin Â3a isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. Human Molecular Genetics, 2003, 12, 2395-2409.	1.4	123
61	Wide spectrum of filaggrin-null mutations in atopic dermatitis highlights differences between Singaporean Chinese and European populations. British Journal of Dermatology, 2011, 165, 106-114.	1.4	123
62	A Homozygous Missense Mutation in TGM5 Abolishes Epidermal Transglutaminase 5 Activity and Causes Acral Peeling Skin Syndrome. American Journal of Human Genetics, 2005, 77, 909-917.	2.6	122
63	A Lack of Premature Termination Codon Read-Through Efficacy of PTC124 (Ataluren) in a Diverse Array of Reporter Assays. PLoS Biology, 2013, 11, e1001593.	2.6	118
64	Filaggrin-stratified transcriptomic analysis of pediatric skin identifies mechanistic pathways in patients with atopic dermatitis. Journal of Allergy and Clinical Immunology, 2014, 134, 82-91.	1.5	118
65	Role of transglutaminase 2 in glucose tolerance: knockout mice studies and a putative mutation in a MODY patient. FASEB Journal, 2002, 16, 1371-1378.	0.2	107
66	Therapeutic siRNAs for dominant genetic skin disorders including pachyonychia congenita. Journal of Dermatological Science, 2008, 51, 151-157.	1.0	107
67	Filaggrin's Fuller Figure: A Glimpse into the Genetic Architecture of Atopic Dermatitis. Journal of Investigative Dermatology, 2007, 127, 1282-1284.	0.3	106
68	The Phenotypic and Molecular Genetic Features of Pachyonychia Congenita. Journal of Investigative Dermatology, 2011, 131, 1015-1017.	0.3	105
69	Keratin disorders: from gene to therapy. Human Molecular Genetics, 2011, 20, R189-R197.	1.4	105
70	Plectin and human genetic disorders of the skin and muscle. The paradigm of epidermolysis bullosa with muscular dystrophy. Experimental Dermatology, 1996, 5, 237-246.	1.4	103
71	Severe dermatitis, multiple allergies, and metabolic wasting syndrome caused by a novel mutation in the N-terminal plakin domain of desmoplakin. Journal of Allergy and Clinical Immunology, 2015, 136, 1268-1276.	1.5	103
72	Ichthyosis Bullosa of Siemensâ€"A Disease Involving Keratin 2e. Journal of Investigative Dermatology, 1994, 103, 277-281.	0.3	99

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73	Single-Nucleotide-Specific siRNA Targeting in a Dominant-Negative Skin Model. Journal of Investigative Dermatology, 2008, 128, 594-605.	0.3	99
74	Clinical and molecular characterization of lipoid proteinosis in Namaqualand, South Africa. British Journal of Dermatology, 2004, 151, 413-423.	1.4	98
75	The Genetic Basis of Pachyonychia Congenita. Journal of Investigative Dermatology Symposium Proceedings, 2005, 10, 21-30.	0.8	98
76	Filaggrin haploinsufficiency is highly penetrant and is associated with increased severity of eczema: further delineation of the skin phenotype in a prospective epidemiological study of 792 school children. British Journal of Dermatology, 2009, 161, 884-889.	1.4	98
77	Frameshift Mutation in the V2 Domain of Human Keratin 1 Results in Striate Palmoplantar Keratoderma. Journal of Investigative Dermatology, 2002, 118, 838-844.	0.3	96
78	Prevalent and Low-Frequency Null Mutations in the Filaggrin Gene Are Associated with Early-Onset and Persistent Atopic Eczema. Journal of Investigative Dermatology, 2008, 128, 1591-1594.	0.3	95
79	Novel filaggrin mutation but no other loss-of-function variants found in Ethiopian patients with atopic dermatitis. British Journal of Dermatology, 2011, 165, 1074-1080.	1.4	95
80	Ichthyosis Bullosa of Siemens Is Caused by Mutations in the Keratin 2e Gene. Journal of Investigative Dermatology, 1994, 103, 286-289.	0.3	94
81	Filaggrin breakdown products determine corneocyte conformation in patients with atopic dermatitis. Journal of Allergy and Clinical Immunology, 2015, 136, 1573-1580.e2.	1.5	93
82	A Novel Connexin 30 Mutation in Clouston Syndrome. Journal of Investigative Dermatology, 2002, 118, 530-532.	0.3	89
83	Keratin 9 Is Required for the Structural Integrity and Terminal Differentiation of the Palmoplantar Epidermis. Journal of Investigative Dermatology, 2014, 134, 754-763.	0.3	87
84	Skin Barrier Function in Healthy Subjects and Patients with Atopic Dermatitis in Relation to Filaggrin Loss-of-Function Mutations. Journal of Investigative Dermatology, 2011, 131, 540-542.	0.3	84
85	Analysis of the individual and aggregate genetic contributions of previously identified serine peptidase inhibitor Kazal type 5 (SPINK5), kallikrein-related peptidase 7 (KLK7), and filaggrin (FLG) polymorphisms to eczema risk. Journal of Allergy and Clinical Immunology, 2008, 122, 560-568.e4.	1.5	83
86	Mutations in AQP5, Encoding a Water-Channel Protein, Cause Autosomal-Dominant Diffuse Nonepidermolytic Palmoplantar Keratoderma. American Journal of Human Genetics, 2013, 93, 330-335.	2.6	82
87	Clumping Factor B Promotes Adherence of Staphylococcus aureus to Corneocytes in Atopic Dermatitis. Infection and Immunity, 2017, 85, .	1.0	79
88	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. Nature Genetics, 2012, 44, 1272-1276.	9.4	78
89	Recurrent Mutations in Kindlin-1, a Novel Keratinocyte Focal Contact Protein, in the Autosomal Recessive Skin Fragility and Photosensitivity Disorder, Kindler Syndrome. Journal of Investigative Dermatology, 2004, 122, 78-83.	0.3	77
90	FLGmutation p.Lys4021X in the C-terminal imperfect filaggrin repeat in Japanese patients with atopic eczema. British Journal of Dermatology, 2009, 161, 1387-1390.	1.4	72

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91	Epithelial Inflammation Resulting from an Inherited Loss-of-Function Mutation in EGFR. Journal of Investigative Dermatology, 2014, 134, 2570-2578.	0.3	71
92	The molecular genetics of the genodermatoses: progress to date and future directions. British Journal of Dermatology, 2003, 148, 1-13.	1.4	70
93	Development of Allele-Specific Therapeutic siRNA for Keratin 5 Mutations in Epidermolysis Bullosa Simplex. Journal of Investigative Dermatology, 2011, 131, 2079-2086.	0.3	70
94	Novel and Recurrent Mutations in the Genes Encoding Keratins K6a, K16 and K17 in 13 Cases of Pachyonychia Congenita. Journal of Investigative Dermatology, 2001, 117, 1391-1396.	0.3	69
95	Filaggrin lossâ€ofâ€function mutations and atopic dermatitis as risk factors for hand eczema in apprentice nurses: part <scp>II</scp> of a prospective cohort study. Contact Dermatitis, 2014, 70, 139-150.	0.8	69
96	K15 Expression Implies Lateral Differentiation within Stratified Epithelial Basal Cells. Laboratory Investigation, 2000, 80, 1701-1710.	1.7	68
97	South African amaXhosa patients with atopic dermatitis have decreased levels of filaggrin breakdown products but no loss-of-function mutations in filaggrin. Journal of Allergy and Clinical Immunology, 2014, 133, 280-282.e2.	1.5	67
98	Copy-Number Mutations on Chromosome 17q24.2-q24.3 in Congenital Generalized Hypertrichosis Terminalis with or without Gingival Hyperplasia. American Journal of Human Genetics, 2009, 84, 807-813.	2.6	66
99	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. American Journal of Human Genetics, 2012, 91, 1115-1121.	2.6	65
100	Development of Therapeutic siRNAs for Pachyonychia Congenita. Journal of Investigative Dermatology, 2008, 128, 50-58.	0.3	64
101	Association between domestic water hardness, chlorine, and atopic dermatitis risk in early life: AÂpopulation-based cross-sectional study. Journal of Allergy and Clinical Immunology, 2016, 138, 509-516.	1.5	64
102	Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies. Human Molecular Genetics, 2018, 27, 1228-1240.	1.4	64
103	Cyclic Ichthyosis with Epidermolytic Hyperkeratosis: A Phenotype Conferred by Mutations in the 2B Domain of Keratin K1. American Journal of Human Genetics, 1999, 64, 732-738.	2.6	62
104	Filaggrin mutations are associated with recurrent skin infection in Singaporean Chinese patients with atopic dermatitis. British Journal of Dermatology, 2012, 166, 200-203.	1.4	62
105	A Large Mutational Study in Pachyonychia Congenita. Journal of Investigative Dermatology, 2011, 131, 1018-1024.	0.3	60
106	Molecular Genetics of Meesmann's Corneal Dystrophy: Ancestral and Novel Mutations in Keratin 12 (K12) and Complete Sequence of the Human KRT12 Gene. Experimental Eye Research, 2000, 70, 41-49.	1.2	58
107	Unique and Recurrent Mutations in the Filaggrin Gene in Singaporean Chinese Patients with Ichthyosis Vulgaris. Journal of Investigative Dermatology, 2008, 128, 1669-1675.	0.3	58
108	Heterozygous Null Alleles in Filaggrin Contribute to Clinical Dry Skin in Young Adults and the Elderly. Journal of Investigative Dermatology, 2009, 129, 1042-1045.	0.3	58

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109	Chromosome 11q13.5 variant associated with childhood eczema: An effect supplementary to filaggrin mutations. Journal of Allergy and Clinical Immunology, 2010, 125, 170-174.e2.	1.5	58
110	Statins Downregulate K6a Promoter Activity: A Possible Therapeutic Avenue for Pachyonychia Congenita. Journal of Investigative Dermatology, 2011, 131, 1045-1052.	0.3	57
111	Novel Molecular Therapies for Heritable Skin Disorders. Journal of Investigative Dermatology, 2012, 132, 820-828.	0.3	57
112	Epidermolysis Bullosa Simplex in Israel. Archives of Dermatology, 2003, 139, 498-505.	1.7	56
113	Epidermolysis bullosa: a spectrum of clinical phenotypes explained by molecular heterogeneity. Trends in Molecular Medicine, 1997, 3, 457-465.	2.6	55
114	Novel Mechanism of Revertant Mosaicism in Dowlingâ€“Meara Epidermolysis Bullosa Simplex. Journal of Investigative Dermatology, 2004, 122, 73-77.	0.3	54
115	Filaggrin gene mutation associations with peanut allergy persist despite variations in peanut allergy diagnostic criteria or asthma status. Journal of Allergy and Clinical Immunology, 2013, 132, 239-242.e7.	1.5	54
116	Filaggrin Mutations Are Genetic Modifying Factors Exacerbating X-Linked Ichthyosis. Journal of Investigative Dermatology, 2007, 127, 2795-2798.	0.3	53
117	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.	0.3	53
118	Filaggrin variants confer susceptibility to asthma. Journal of Allergy and Clinical Immunology, 2008, 121, 1294-1295.	1.5	52
119	Lysosomal protease deficiency or substrate overload induces an oxidative-stress mediated STAT3-dependent pathway of lysosomal homeostasis. Nature Communications, 2018, 9, 5343.	5.8	52
120	Novel keratin 16 mutations and protein expression studies in pachyonychia congenita type 1 and focal palmoplantar keratoderma. Experimental Dermatology, 2000, 9, 170-177.	1.4	51
121	Missing Câ€terminal filaggrin expression, NFkappaB activation and hyperproliferation identify the dog as a putative model to study epidermal dysfunction in atopic dermatitis. Experimental Dermatology, 2010, 19, e343-6.	1.4	51
122	A novel mutation in the helix termination motif of keratin K12 in a US family with Meesmann corneal dystrophy. American Journal of Ophthalmology, 1999, 128, 687-691.	1.7	50
123	Defolliculated (Dfl): A Dominant Mouse Mutation Leading to Poor Sebaceous Gland Differentiation and Total Elimination of Pelage Follicles. Journal of Investigative Dermatology, 2002, 119, 32-37.	0.3	50
124	Keratin K6c Mutations Cause Focal Palmoplantar Keratoderma. Journal of Investigative Dermatology, 2010, 130, 425-429.	0.3	50
125	Comprehensive screening for a complete set of Japaneseâ€populationâ€specific filaggrin gene mutations. Allergy: European Journal of Allergy and Clinical Immunology, 2014, 69, 537-540.	2.7	50
126	The Gene for Hypotrichosis of Marie Unna Maps between D8S258 and D8S298: Exclusion of the hr Gene by cDNA and Genomic Sequencing. American Journal of Human Genetics, 1999, 65, 413-419.	2.6	49

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127	Two Cases of Primarily Palmoplantar Keratoderma Associated with Novel Mutations in Keratin 1. Journal of Investigative Dermatology, 2002, 119, 966-971.	0.3	49
128	A spectrum of mutations in keratins K6a, K16 and K17 causing pachyonychia congenita. Journal of Dermatological Science, 2007, 48, 199-205.	1.0	49
129	Analysis of Taiwanese ichthyosis vulgaris families further demonstrates differences inFLGmutations between European and Asian populations. British Journal of Dermatology, 2009, 161, 448-451.	1.4	49
130	Clouston Syndrome Can Mimic Pachyonychia Congenita. Journal of Investigative Dermatology, 2003, 121, 1035-1038.	0.3	48
131	Atypical epidermolytic palmoplantar keratoderma presentation associated with a mutation in the keratin 1 gene. British Journal of Dermatology, 2004, 150, 1096-1103.	1.4	48
132	Mutations in GRHL2 Result in an Autosomal-Recessive Ectodermal Dysplasia Syndrome. American Journal of Human Genetics, 2014, 95, 308-314.	2.6	48
133	Mutations in Keratin K9 in Kindreds with Epidermolytic Palmoplantar Keratoderma and Epidemiology in Northern Ireland. Journal of Investigative Dermatology, 1998, 111, 1207-1209.	0.3	47
134	A Mutation in the V1 Domain of K16 is Responsible for Unilateral Palmoplantar Verrucous Nevus. Journal of Investigative Dermatology, 2000, 114, 1136-1140.	0.3	47
135	A Heterozygous Frameshift Mutation in the V1 Domain of Keratin 5 in a Family with Dowlingâ€“Degos Disease. Journal of Investigative Dermatology, 2007, 127, 298-300.	0.3	47
136	Filaggrin null mutations are associated with increased asthma exacerbations in children and young adults. Allergy: European Journal of Allergy and Clinical Immunology, 2008, 63, 1211-1217.	2.7	46
137	cDNA Cloning, mRNA Expression, and Chromosomal Mapping of Human and Mouse Periplakin Genes. Genomics, 1998, 48, 242-247.	1.3	45
138	Prevalent and Rare Mutations in the Gene Encoding Filaggrin in Japanese Patients with Ichthyosis Vulgaris and Atopic Dermatitis. Journal of Investigative Dermatology, 2009, 129, 1302-1305.	0.3	43
139	Japanese-Specific Filaggrin Gene Mutations in Japanese Patients Suffering from Atopic Eczema and Asthma. Journal of Investigative Dermatology, 2010, 130, 2834-2836.	0.3	43
140	Gut microbiota development during infancy: Impact of introducing allergenic foods. Journal of Allergy and Clinical Immunology, 2021, 147, 613-621.e9.	1.5	43
141	Delayed-onset pachyonychia congenita associated with a novel mutation in the central 2B domain of keratin 16. British Journal of Dermatology, 2001, 144, 1058-1062.	1.4	42
142	Expanding the clinical spectrum of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis due to FAM111B mutations. Orphanet Journal of Rare Diseases, 2015, 10, 135.	1.2	42
143	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex. , 1998, 11, 279-285.		41
144	Filaggrin Null Alleles Are Not Associated with Psoriasis. Journal of Investigative Dermatology, 2007, 127, 1878-1882.	0.3	41

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145	De Novo Occurrence of the Filaggrin Mutation p.R501X with Prevalent Mutation c.3321delA in a Japanese Family with Ichthyosis Vulgaris Complicated by Atopic Dermatitis. Journal of Investigative Dermatology, 2008, 128, 1323-1325.	0.3	40
146	Ichthyosis vulgaris: novelFLGmutations in the German population and high presence of CD1a+ cells in the epidermis of the atopic subgroup. British Journal of Dermatology, 2009, 160, 771-781.	1.4	40
147	Insights into Genotypeâ€“Phenotype Correlation in Pachyonychia Congenita from the Human Intermediate Filament Mutation Database. Journal of Investigative Dermatology Symposium Proceedings, 2005, 10, 31-36.	0.8	39
148	Cloning of multiple keratin 16 genes facilitates prenatal diagnosis of pachyonychia congenita type 1. , 1999, 19, 941-946.		38
149	Generic and Personalized RNAi-Based Therapeutics for a Dominant-Negative Epidermal Fragility Disorder. Journal of Investigative Dermatology, 2012, 132, 1627-1635.	0.3	38
150	Development of Allele-Specific Therapeutic siRNA in Meesmann Epithelial Corneal Dystrophy. PLoS ONE, 2011, 6, e28582.	1.1	37
151	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.	0.3	37
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