W H Irwin Mclean

List of Publications by Year
in descending order

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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	Gut microbiota development during infancy: Impact of introducing allergenic foods. Journal of Allergy and Clinical Immunology, 2021, 147, 613-621.e9.	1.5	43
2	Male genital lichen sclerosus and filaggrin. Clinical and Experimental Dermatology, 2020, 45, 127-128.	0.6	3
3	Bathing frequency is associated with skin barrier dysfunction and atopic dermatitis at three months of age. Journal of Allergy and Clinical Immunology: in Practice, 2020, 8, 2820-2822.	2.0	21
4	Development of a Corneal Bioluminescence Mouse for Real-Time In Vivo Evaluation of Gene Therapies. Translational Vision Science and Technology, 2020, 9, 44.	1.1	2
5	Filaggrin gene lossâ€ofâ€function mutations constitute a factor in patients with multiple contact allergies. Contact Dermatitis, 2019, 80, 354-358.	0.8	15
6	Discovery of Soft-Drug Topical Tool Modulators of Sphingosine-1-phosphate Receptor 1 (S1PR1). ACS Medicinal Chemistry Letters, 2019, 10, 341-347.	1.3	5
7	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
8	Gentamicin-Induced Readthrough and Nonsense-Mediated mRNA Decay of SERPINB7 Nonsense Mutant Transcripts. Journal of Investigative Dermatology, 2018, 138, 836-843.	0.3	33
9	Array-based sequencing of filaggrin gene for comprehensive detection of disease-associated variants. Journal of Allergy and Clinical Immunology, 2018, 141, 814-816.	1.5	36
10	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
11	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
12	Discovery of super soft-drug modulators of sphingosine-1-phosphate receptor 1. Bioorganic and Medicinal Chemistry Letters, 2018, 28, 3255-3259.	1.0	18
13	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. American Journal of Human Genetics, 2017, 100, 364-370.	2.6	32
14	Clumping Factor B Promotes Adherence of Staphylococcus aureus to Corneocytes in Atopic Dermatitis. Infection and Immunity, 2017, 85, .	1.0	79
15	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
16	Calpain 12 Function Revealed through the Study of an Atypical Case of Autosomal Recessive Congenital Ichthyosis. Journal of Investigative Dermatology, 2017, 137, 385-393.	0.3	19
17	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
18	Patients with atopic dermatitis with filaggrin loss-of-function mutations show good but lower responses to immunosuppressive treatment. British Journal of Dermatology, 2017, 177, 1745-1746.	1.4	11

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19	Keratin 6b variant p.Gly499Ser reported in delayedâ€onset pachyonychia congenita is a nonâ€pathogenic polymorphism. Journal of Dermatology, 2017, 44, e312.	0.6	3
20	A novel keratin 13 variant in a fourâ€generation family with white sponge nevus. Clinical Case Reports (discontinued), 2017, 5, 1503-1509.	0.2	12
21	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
22	Filaggrin genotype does not determine the skin's threshold to UV-induced erythema. Journal of Allergy and Clinical Immunology, 2016, 137, 1280-1282.e3.	1.5	6
23	Keratin 12 missense mutation induces the unfolded protein response and apoptosis in Meesmann epithelial corneal dystrophy. Human Molecular Genetics, 2016, 25, 1176-1191.	1.4	22
24	Filaggrin-null mutations are associated with increased maturation markers on Langerhans cells. Journal of Allergy and Clinical Immunology, 2016, 138, 482-490.e7.	1.5	28
25	Too Much, Too Little or Just Enough: A Goldilocks Effect for IL-13 and Skin Barrier Regulation?. Journal of Investigative Dermatology, 2016, 136, 561-564.	0.3	16
26	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
27	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
28	Novel <scp>TGM</scp>5 mutations in acral peeling skin syndrome. Experimental Dermatology, 2015, 24, 285-289.	1.4	11
29	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
30	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
31	Loss-of-Function Mutations in CAST Cause Peeling Skin, Leukonychia, Acral Punctate Keratoses, Cheilitis, and Knuckle Pads. American Journal of Human Genetics, 2015, 96, 440-447.	2.6	36
32	Loss-of-Function Mutations in the Gene Encoding Filaggrin Are Not Strongly Associated with Chronic Actinic Dermatitis. Journal of Investigative Dermatology, 2015, 135, 1919-1921.	0.3	6
33	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
34	Punctate Palmoplantar Keratoderma Type 1: A Novel AAGAB Mutation and Efficacy of Etretinate. Acta Dermato-Venereologica, 2015, 95, 110-111.	0.6	14
35	PCQoL. Journal of Cutaneous Medicine and Surgery, 2015, 19, 57-65.	0.6	6
36	Expanding the Phenotypic Spectrum of Olmsted Syndrome. Journal of Investigative Dermatology, 2015, 135, 2879-2883.	0.3	23

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37	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
38	Lysyl Hydroxylase 3 Localizes to Epidermal Basement Membrane and Is Reduced in Patients with Recessive Dystrophic Epidermolysis Bullosa. PLoS ONE, 2015, 10, e0137639.	1.1	21
39	Improved Annotation of 3â€² Untranslated Regions and Complex Loci by Combination of Strand-Specific Direct RNA Sequencing, RNA-Seq and ESTs. PLoS ONE, 2014, 9, e94270.	1.1	27
40	A new filaggrin gene mutation in a Korean patient with ichthyosis vulgaris. European Journal of Dermatology, 2014, 24, 491-493.	0.3	2
41	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
42	Mutations in GRHL2 Result in an Autosomal-Recessive Ectodermal Dysplasia Syndrome. American Journal of Human Genetics, 2014, 95, 308-314.	2.6	48
43	Plectin Mutations Underlie Epidermolysis Bullosa Simplex in 8% of Patients. Journal of Investigative Dermatology, 2014, 134, 273-276.	0.3	34
44	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
45	In vivo gene silencing following non-invasive siRNA delivery into the skin using a novel topical formulation. Journal of Controlled Release, 2014, 196, 355-362.	4.8	34
46	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
47	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
48	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
49	siRNA Silencing of the Mutant Keratin 12 Allele in Corneal Limbal Epithelial Cells Grown From Patients With Meesmann's Epithelial Corneal Dystrophy. , 2014, 55, 3352.		28
50	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
51	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
52	Epithelial Inflammation Resulting from an Inherited Loss-of-Function Mutation in EGFR. Journal of Investigative Dermatology, 2014, 134, 2570-2578.	0.3	71
53	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
54	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

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55	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
56	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
57	Recessive mutations in the gene encoding frizzled 6 cause twenty nail dystrophyâ€"Expanding the differential diagnosis for pachyonychia congenita. Journal of Dermatological Science, 2013, 70, 58-60.	1.0	22
58	Reliability and validity of genotyping filaggrin null mutations. Journal of Dermatological Science, 2013, 70, 67-68.	1.0	9
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	Possibilities for human skin characterization based on strongly reduced Raman spectroscopic information. Journal of Raman Spectroscopy, 2013, 44, 340-345.	1.2	12
61	High-density genotyping study identifies four new susceptibility loci for atopic dermatitis. Nature Genetics, 2013, 45, 808-812.	9.4	167
62	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
63	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
64	Resolution of the plantar hyperkeratosis of pachyonychia congenita during chemotherapy for Ewing sarcoma. British Journal of Dermatology, 2013, 169, 1357-1360.	1.4	3
65	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. Nature Genetics, 2013, 45, 1244-1248.	9.4	289
66	Heterozygous Mutations in AAGAB Cause Type 1 Punctate Palmoplantar Keratoderma with Evidence for Increased Growth Factor Signaling. Journal of Investigative Dermatology, 2013, 133, 2805-2808.	0.3	21
67	Generation and Characterisation of Keratin 7 (K7) Knockout Mice. PLoS ONE, 2013, 8, e64404.	1.1	35
68	Allele-Specific siRNA Silencing for the Common Keratin 12 Founder Mutation in Meesmann Epithelial Corneal Dystrophy. , 2013, 54, 494.		34
69	Old King Coal â€" molecular mechanisms underlying an ancient treatment for atopic eczema. Journal of Clinical Investigation, 2013, 123, 551-3.	3.9	11
70	Mutations in the SASPase Gene (ASPRV1) Are Not Associated with Atopic Eczema or Clinically Dry Skin. Journal of Investigative Dermatology, 2012, 132, 1507-1510.	0.3	10
71	Novel Molecular Therapies for Heritable Skin Disorders. Journal of Investigative Dermatology, 2012, 132, 820-828.	0.3	57
72	Heritable Filaggrin Disorders: The Paradigm of Atopic Dermatitis. Journal of Investigative Dermatology, 2012, 132, E20-E21.	0.3	22

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73	Generic and Personalized RNAi-Based Therapeutics for a Dominant-Negative Epidermal Fragility Disorder. Journal of Investigative Dermatology, 2012, 132, 1627-1635.	0.3	38
74	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. Nature Genetics, 2012, 44, 1272-1276.	9.4	78
75	One Remarkable Molecule: Filaggrin. Journal of Investigative Dermatology, 2012, 132, 751-762.	0.3	433
76	Homozygous Dominant Missense Mutation in Keratin 17 Leads to Alopecia in Addition to Severe Pachyonychia Congenita. Journal of Investigative Dermatology, 2012, 132, 1921-1924.	0.3	20
77	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
78	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
79	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
80	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
81	A recurrent mutation in the TGM5 gene in European patients with acral peeling skin syndrome. Journal of Dermatological Science, 2012, 65, 74-76.	1.0	10
82	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
83	Transgrediens pachyonychia congenita (PC): case series of a nonclassical PC presentation. British Journal of Dermatology, 2012, 166, 124-128.	1.4	6
84	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
85	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
86	Exacerbation of X-linked ichthyosis phenotype in a female by inheritance of filaggrin and steroid sulfatase mutations. Journal of Dermatological Science, 2011, 64, 159-162.	1.0	25
87	Filaggrin Mutations Associated with Skin and Allergic Diseases. New England Journal of Medicine, 2011, 365, 1315-1327.	13.9	996
88	Filaggrin Genotype in Ichthyosis Vulgaris Predicts Abnormalities in Epidermal Structure and Function. American Journal of Pathology, 2011, 178, 2252-2263.	1.9	213
89	The Phenotypic and Molecular Genetic Features of Pachyonychia Congenita. Journal of Investigative Dermatology, 2011, 131, 1015-1017.	0.3	105
90	A Large Mutational Study in Pachyonychia Congenita. Journal of Investigative Dermatology, 2011, 131, 1018-1024.	0.3	60

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91	Development of Allele-Specific Therapeutic siRNA in Meesmann Epithelial Corneal Dystrophy. PLoS ONE, 2011, 6, e28582.	1.1	37
92	Paternal Germ Cell Mosaicism in Autosomal Dominant Pachyonychia Congenita. Archives of Dermatology, 2011, 147, 1077.	1.7	4
93	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
94	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
95	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
96	Identification of a novel C16orf57 mutation in Athabaskan patients with Poikiloderma with Neutropenia. American Journal of Medical Genetics, Part A, 2011, 155, 337-342.	0.7	34
97	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
98	Keratin disorders: from gene to therapy. Human Molecular Genetics, 2011, 20, R189-R197.	1.4	105
99	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
100	Development of Quantitative Molecular Clinical End Points for siRNA Clinical Trials. Journal of Investigative Dermatology, 2011, 131, 1029-1036.	0.3	21
101	Obtaining DNA in the Mail from a National Sample of Children with a Chronic Non-Fatal Illness. Journal of Investigative Dermatology, 2011, 131, 1765-1767.	0.3	9
102	Statins Downregulate K6a Promoter Activity: A Possible Therapeutic Avenue for Pachyonychia Congenita. Journal of Investigative Dermatology, 2011, 131, 1045-1052.	0.3	57
103	The allergy gene: how a mutation in a skin protein revealed a link between eczema and asthma. F1000 Medicine Reports, 2011, 3, 2.	2.9	14
104	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
105	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
106	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
107	Keratin K6c Mutations Cause Focal Palmoplantar Keratoderma. Journal of Investigative Dermatology, 2010, 130, 425-429.	0.3	50
108	Deletion of Late Cornified Envelope 3B and 3C Genes Is Not Associated with Atopic Dermatitis. Journal of Investigative Dermatology, 2010, 130, 2057-2061.	0.3	25

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109	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
110	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
111	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
112	Chromosome 11q13.5 variant: No association with atopic eczema in the Japanese population. Journal of Dermatological Science, 2010, 59, 210-212.	1.0	2
113	Carrier Status for the Common R501X and 2282del4 Filaggrin Mutations Is Not Associated with Hearing Phenotypes in 5377 Children from the ALSPAC Cohort. PLoS ONE, 2009, 4, e5784.	1.1	8
114	Keratin 7 promoter selectively targets transgene expression to normal and neoplastic pancreatic ductal cellsin vitroandin vivo. FASEB Journal, 2009, 23, 1366-1375.	0.2	17
115	Identification of a Novel Family of Laminin N-terminal Alternate Splice Isoforms. Journal of Biological Chemistry, 2009, 284, 35588-35596.	1.6	26
116	Filaggrin loss-of-function variants are associated with atopic comorbidity in pediatric inflammatory bowel disease. Inflammatory Bowel Diseases, 2009, 15, 1492-1498.	0.9	22
117	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
118	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
119	Mutation inDSG1causing autosomal dominant striate palmoplantar keratoderma. British Journal of Dermatology, 2009, 161, 692-694.	1.4	17
120	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
121	Steatocystoma multiplex, oligodontia and partial persistent primary dentition associated with a novel keratin 17 mutation. British Journal of Dermatology, 2009, 161, 1396-1398.	1.4	19
122	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
123	Association of Skin Barrier Genes within the PSORS4 Locus Is Enriched in Singaporean Chinese with Early-Onset Psoriasis. Journal of Investigative Dermatology, 2009, 129, 606-614.	0.3	23
124	Clinical Severity Correlates with Impaired Barrier in Filaggrin-Related Eczema. Journal of Investigative Dermatology, 2009, 129, 682-689.	0.3	154
125	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
126	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

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127	Eczema Genetics: Current State of Knowledge and Future Goals. Journal of Investigative Dermatology, 2009, 129, 543-552.	0.3	139
128	Achieving Successful Delivery of Nucleic Acids to Skin: 6th Annual Meeting of the International Pachyonychia Congenita Consortium. Journal of Investigative Dermatology, 2009, 129, 2085-2087.	0.3	13
129	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
130	A homozygous frameshift mutation in the mouse Flg gene facilitates enhanced percutaneous allergen priming. Nature Genetics, 2009, 41, 602-608.	9.4	438
131	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
132	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
133	Filaggrin in atopic dermatitis. Journal of Allergy and Clinical Immunology, 2009, 124, R2-R6.	1.5	245
134	Filaggrin in the frontline: role in skin barrier function and disease. Journal of Cell Science, 2009, 122, 1285-1294.	1.2	672
135	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
136	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
137	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
138	Combing the genome for the root cause of baldness. Nature Genetics, 2008, 40, 1270-1271.	9.4	7
139	Development of Therapeutic siRNAs for Pachyonychia Congenita. Journal of Investigative Dermatology, 2008, 128, 50-58.	0.3	64
140	Single-Nucleotide-Specific siRNA Targeting in a Dominant-Negative Skin Model. Journal of Investigative Dermatology, 2008, 128, 594-605.	0.3	99
141	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
142	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
143	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
144	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

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145	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
146	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
147	Filaggrin mutations, atopic eczema, hay fever, and asthma in children. Journal of Allergy and Clinical Immunology, 2008, 121, 1203-1209.e1.	1.5	380
148	Filaggrin variants confer susceptibility to asthma. Journal of Allergy and Clinical Immunology, 2008, 121, 1294-1295.	1.5	52
149	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
150	Filaggrin in atopic dermatitis. Journal of Allergy and Clinical Immunology, 2008, 122, 689-693.	1.5	410
151	Therapeutic siRNAs for dominant genetic skin disorders including pachyonychia congenita. Journal of Dermatological Science, 2008, 51, 151-157.	1.0	107
152	Broken bricks and cracked mortar â€“ epidermal diseases resulting from genetic abnormalities. Drug Discovery Today Disease Mechanisms, 2008, 5, e93-e101.	0.8	6
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