W H Irwin Mclean

List of Publications by Year in Descending Order

249 papers	23,536 citations	79 h-index	148 g-index
257 ext. papers	26,276 ext. citations	5.6 avg, IF	6.42 L-index

#	Paper	IF	Citations
249	Gut microbiota development during infancy: Impact of introducing allergenic foods. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 613-621.e9	11.5	10
248	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	5.4	8
247	Development of a Corneal Bioluminescence Mouse for Real-Time In Vivo Evaluation of Gene Therapies. *Translational Vision Science and Technology*, **2020**, 9, 44	3.3	1
246	Male genital lichen sclerosus and filaggrin. *Clinical and Experimental Dermatology*, **2020**, 45, 127-128	1.8	3
245	Filaggrin gene loss-of-function mutations constitute a factor in patients with multiple contact allergies. *Contact Dermatitis*, **2019**, 80, 354-358	2.7	9
244	Discovery of Soft-Drug Topical Tool Modulators of Sphingosine-1-phosphate Receptor 1 (S1PR1). *ACS Medicinal Chemistry Letters*, **2019**, 10, 341-347	4.3	3
243	Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies. *Human Molecular Genetics*, **2018**, 27, 1228-1240	5.6	40
242	Gentamicin-Induced Readthrough and Nonsense-Mediated mRNA Decay of SERPINB7 Nonsense Mutant Transcripts. *Journal of Investigative Dermatology*, **2018**, 138, 836-843	4.3	17
241	Array-based sequencing of filaggrin gene for comprehensive detection of disease-associated variants. *Journal of Allergy and Clinical Immunology*, **2018**, 141, 814-816	11.5	26
240	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
239	Discovery of super soft-drug modulators of sphingosine-1-phosphate receptor 1. *Bioorganic and Medicinal Chemistry Letters*, **2018**, 28, 3255-3259	2.9	13
238	Lysosomal protease deficiency or substrate overload induces an oxidative-stress mediated STAT3-dependent pathway of lysosomal homeostasis. *Nature Communications*, **2018**, 9, 5343	17.4	33
237	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. *American Journal of Human Genetics*, **2017**, 100, 364-370	11	22
236	Clumping Factor B Promotes Adherence of Staphylococcus aureus to Corneocytes in Atopic Dermatitis. *Infection and Immunity*, **2017**, 85,	3.7	53
235	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
234	Calpain 12 Function Revealed through the Study of an Atypical Case of Autosomal Recessive Congenital Ichthyosis. *Journal of Investigative Dermatology*, **2017**, 137, 385-393	4.3	14
233	Skin microbiome before development of atopic dermatitis: Early colonization with commensal staphylococci at 2 months is associated with a lower risk of atopic dermatitis at 1 year. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 166-172	11.5	186

232	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	4	6
231	Keratin 6b variant p.Gly499Ser reported in delayed-onset pachyonychia congenita is a non-pathogenic polymorphism. *Journal of Dermatology*, **2017**, 44, e312	1.6	2
230	A novel keratin 13 variant in a four-generation family with white sponge nevus. *Clinical Case Reports (discontinued)*, **2017**, 5, 1503-1509	0.7	7
229	Filaggrin genotype does not determine the skin threshold to UV-induced erythema. *Journal of Allergy and Clinical Immunology*, **2016**, 137, 1280-1282.e3	11.5	6
228	Keratin 12 missense mutation induces the unfolded protein response and apoptosis in Meesmann epithelial corneal dystrophy. *Human Molecular Genetics*, **2016**, 25, 1176-91	5.6	11
227	Filaggrin-null mutations are associated with increased maturation markers on Langerhans cells. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 482-490.e7	11.5	18
226	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	4.3	10
225	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	11.5	108
224	Association between domestic water hardness, chlorine, and atopic dermatitis risk in early life: A population-based cross-sectional study. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 509-16	11.5	48
223	Skin barrier impairment at birth predicts food allergy at 2 years of age. *Journal of Allergy and Clinical Immunology*, **2016**, 137, 1111-1116.e8	11.5	133
222	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	4.3	3
221	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, 1268-76	11.5	82
220	Punctate palmoplantar keratoderma type 1: a novel AAGAB mutation and efficacy of etretinate. *Acta Dermato-Venereologica*, **2015**, 95, 110-1	2.2	8
219	PCQoL: A Quality of Life Assessment Measure for Pachyonychia Congenita. *Journal of Cutaneous Medicine and Surgery*, **2015**, 19, 57-65	1.6	3
218	Skin barrier dysfunction measured by transepidermal water loss at 2 days and 2 months predates and predicts atopic dermatitis at 1 year. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 930-935.e1	11.5	180
217	Expanding the Phenotypic Spectrum of Olmsted Syndrome. *Journal of Investigative Dermatology*, **2015**, 135, 2879-2883	4.3	19
216	Filaggrin breakdown products determine corneocyte conformation in patients with atopic dermatitis. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 1573-1580.e2	11.5	60
215	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	4.2	17

214	Novel TGM5 mutations in acral peeling skin syndrome. *Experimental Dermatology*, **2015**, 24, 285-9	4	10
213	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, 164-70	11.5	211
212	Genome-wide comparative analysis of atopic dermatitis and psoriasis gives insight into opposing genetic mechanisms. *American Journal of Human Genetics*, **2015**, 96, 104-20	11	113
211	Loss-of-function mutations in CAST cause peeling skin, leukonychia, acral punctate keratoses, cheilitis, and knuckle pads. *American Journal of Human Genetics*, **2015**, 96, 440-7	11	22
210	Lysyl Hydroxylase 3 Localizes to Epidermal Basement Membrane and Is Reduced in Patients with Recessive Dystrophic Epidermolysis Bullosa. *PLoS ONE*, **2015**, 10, e0137639	3.7	18
209	Comprehensive screening for a complete set of Japanese-population-specific filaggrin gene mutations. *Allergy: European Journal of Allergy and Clinical Immunology*, **2014**, 69, 537-40	9.3	37
208	siRNA silencing of the mutant keratin 12 allele in corneal limbal epithelial cells grown from patients with Meesmann epithelial corneal dystrophy **2014**, 55, 3352-60		21
207	Emollient enhancement of the skin barrier from birth offers effective atopic dermatitis prevention. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 818-23	11.5	461
206	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	11.5	186
205	Epithelial inflammation resulting from an inherited loss-of-function mutation in EGFR. *Journal of Investigative Dermatology*, **2014**, 134, 2570-2578	4.3	51
204	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	11.5	89
203	Improved annotation of 3untranslated regions and complex loci by combination of strand-specific direct RNA sequencing, RNA-Seq and ESTs. *PLoS ONE*, **2014**, 9, e94270	3.7	21
202	A new filaggrin gene mutation in a Korean patient with ichthyosis vulgaris. *European Journal of Dermatology*, **2014**, 24, 491-3	0.8	2
201	Keratin 9 is required for the structural integrity and terminal differentiation of the palmoplantar epidermis. *Journal of Investigative Dermatology*, **2014**, 134, 754-763	4.3	55
200	Mutations in GRHL2 result in an autosomal-recessive ectodermal Dysplasia syndrome. *American Journal of Human Genetics*, **2014**, 95, 308-14	11	37
199	Plectin mutations underlie epidermolysis bullosa simplex in 8% of patients. *Journal of Investigative Dermatology*, **2014**, 134, 273-276	4.3	26
198	Filaggrin loss-of-function mutations and atopic dermatitis as risk factors for hand eczema in apprentice nurses: part II of a prospective cohort study. *Contact Dermatitis*, **2014**, 70, 139-50	2.7	59
197	In vivo gene silencing following non-invasive siRNA delivery into the skin using a novel topical formulation. *Journal of Controlled Release*, **2014**, 196, 355-62	11.7	34

196	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, 280-2.e1-2	11.5	58
195	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	4.3	118
194	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, 1121-9	11.5	108
193	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, 239-42	11.5	42
192	Mutations in AQP5, encoding a water-channel protein, cause autosomal-dominant diffuse nonepidermolytic palmoplantar keratoderma. *American Journal of Human Genetics*, **2013**, 93, 330-5	11	61
191	Recessive mutations in the gene encoding frizzled 6 cause twenty nail dystrophy--expanding the differential diagnosis for pachyonychia congenita. *Journal of Dermatological Science*, **2013**, 70, 58-60	4.3	16
190	Reliability and validity of genotyping filaggrin null mutations. *Journal of Dermatological Science*, **2013**, 70, 67-8	4.3	8
189	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	4	95
188	Possibilities for human skin characterization based on strongly reduced Raman spectroscopic information. *Journal of Raman Spectroscopy*, **2013**, 44, 340-345	2.3	10
187	High-density genotyping study identifies four new susceptibility loci for atopic dermatitis. *Nature Genetics*, **2013**, 45, 808-12	36.3	131
186	A lack of premature termination codon read-through efficacy of PTC124 (Ataluren) in a diverse array of reporter assays. *PLoS Biology*, **2013**, 11, e1001593	9.7	96
185	A genome-wide association study of atopic dermatitis identifies loci with overlapping effects on asthma and psoriasis. *Human Molecular Genetics*, **2013**, 22, 4841-56	5.6	140
184	Resolution of the plantar hyperkeratosis of pachyonychia congenita during chemotherapy for Ewing sarcoma. *British Journal of Dermatology*, **2013**, 169, 1357-60	4	3
183	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. *Nature Genetics*, **2013**, 45, 1244-1248	36.3	217
182	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	4.3	11
181	Generation and characterisation of keratin 7 (K7) knockout mice. *PLoS ONE*, **2013**, 8, e64404	3.7	24
180	Allele-specific siRNA silencing for the common keratin 12 founder mutation in Meesmann epithelial corneal dystrophy **2013**, 54, 494-502		29
179	Old King coal - molecular mechanisms underlying an ancient treatment for atopic eczema. *Journal of Clinical Investigation*, **2013**, 123, 551-3	15.9	6

178	One remarkable molecule: filaggrin. *Journal of Investigative Dermatology*, **2012**, 132, 751-62	4.3	343
177	Homozygous dominant missense mutation in keratin 17 leads to alopecia in addition to severe pachyonychia congenita. *Journal of Investigative Dermatology*, **2012**, 132, 1921-4	4.3	13
176	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	4.3	148
175	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nature Genetics*, **2012**, 44, 1341-8	36.3	681
174	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, 1031-9.e1	11.5	179
173	The persistence of atopic dermatitis and filaggrin (FLG) mutations in a US longitudinal cohort. *Journal of Allergy and Clinical Immunology*, **2012**, 130, 912-7	11.5	145
172	A recurrent mutation in the TGM5 gene in European patients with acral peeling skin syndrome. *Journal of Dermatological Science*, **2012**, 65, 74-6	4.3	8
171	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
170	Transgrediens pachyonychia congenita (PC): case series of a nonclassical PC presentation. *British Journal of Dermatology*, **2012**, 166, 124-8	4	3
169	Filaggrin mutations are associated with recurrent skin infection in Singaporean Chinese patients with atopic dermatitis. *British Journal of Dermatology*, **2012**, 166, 200-3	4	49
168	Mutations in the SASPase gene (ASPRV1) are not associated with atopic eczema or clinically dry skin. *Journal of Investigative Dermatology*, **2012**, 132, 1507-10	4.3	8
167	Novel molecular therapies for heritable skin disorders. *Journal of Investigative Dermatology*, **2012**, 132, 820-8	4.3	49
166	Heritable filaggrin disorders: the paradigm of atopic dermatitis. *Journal of Investigative Dermatology*, **2012**, 132, E20-1	4.3	18
165	Generic and personalized RNAi-based therapeutics for a dominant-negative epidermal fragility disorder. *Journal of Investigative Dermatology*, **2012**, 132, 1627-35	4.3	34
164	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. *Nature Genetics*, **2012**, 44, 1272-6	36.3	56
163	Loss-of-function variants in the filaggrin gene are a significant risk factor for peanut allergy. *Journal of Allergy and Clinical Immunology*, **2011**, 127, 661-7	11.5	342
162	Exacerbation of X-linked ichthyosis phenotype in a female by inheritance of filaggrin and steroid sulfatase mutations. *Journal of Dermatological Science*, **2011**, 64, 159-62	4.3	20
161	Filaggrin mutations associated with skin and allergic diseases. *New England Journal of Medicine*, **2011**, 365, 1315-27	59.2	803

160	Genodermatoses: Inherited Diseases of the Skin **2011**, 379-409		1
159	Filaggrin genotype in ichthyosis vulgaris predicts abnormalities in epidermal structure and function. *American Journal of Pathology*, **2011**, 178, 2252-63	5.8	182
158	The phenotypic and molecular genetic features of pachyonychia congenita. *Journal of Investigative Dermatology*, **2011**, 131, 1015-7	4.3	84
157	A large mutational study in pachyonychia congenita. *Journal of Investigative Dermatology*, **2011**, 131, 1018-24	4.3	43
156	Development of allele-specific therapeutic siRNA in Meesmann epithelial corneal dystrophy. *PLoS ONE*, **2011**, 6, e28582	3.7	30
155	Paternal germ cell mosaicism in autosomal dominant pachyonychia congenita. *Archives of Dermatology*, **2011**, 147, 1077-80		4
154	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, 934-40	9.3	207
153	Wide spectrum of filaggrin-null mutations in atopic dermatitis highlights differences between Singaporean Chinese and European populations. *British Journal of Dermatology*, **2011**, 165, 106-14	4	99
152	Novel filaggrin mutation but no other loss-of-function variants found in Ethiopian patients with atopic dermatitis. *British Journal of Dermatology*, **2011**, 165, 1074-80	4	70
151	Identification of a novel C16orf57 mutation in Athabaskan patients with Poikiloderma with Neutropenia. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 337-42	2.5	31
150	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, 540-2	4.3	67
149	Keratin disorders: from gene to therapy. *Human Molecular Genetics*, **2011**, 20, R189-97	5.6	83
148	Development of allele-specific therapeutic siRNA for keratin 5 mutations in epidermolysis bullosa simplex. *Journal of Investigative Dermatology*, **2011**, 131, 2079-86	4.3	53
147	Development of quantitative molecular clinical end points for siRNA clinical trials. *Journal of Investigative Dermatology*, **2011**, 131, 1029-36	4.3	17
146	Obtaining DNA in the mail from a national sample of children with a chronic non-fatal illness. *Journal of Investigative Dermatology*, **2011**, 131, 1765-7	4.3	8
145	Statins downregulate K6a promoter activity: a possible therapeutic avenue for pachyonychia congenita. *Journal of Investigative Dermatology*, **2011**, 131, 1045-52	4.3	46
144	The allergy gene: how a mutation in a skin protein revealed a link between eczema and asthma. *F1000 Medicine Reports*, **2011**, 3, 2		11
143	Missing C-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	4	44

142	Filaggrin loss-of-function mutations are associated with early-onset eczema, eczema severity and transepidermal water loss at 3 months of age. *British Journal of Dermatology*, **2010**, 163, 1333-6	4	164
141	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. *Nature Genetics*, **2010**, 42, 985-90	36.3	773
140	Keratin K6c mutations cause focal palmoplantar keratoderma. *Journal of Investigative Dermatology*, **2010**, 130, 425-9	4.3	37
139	Deletion of Late Cornified Envelope 3B and 3C genes is not associated with atopic dermatitis. *Journal of Investigative Dermatology*, **2010**, 130, 2057-61	4.3	24
138	Japanese-specific filaggrin gene mutations in Japanese patients suffering from atopic eczema and asthma. *Journal of Investigative Dermatology*, **2010**, 130, 2834-6	4.3	31
137	Chromosome 11q13.5 variant associated with childhood eczema: an effect supplementary to filaggrin mutations. *Journal of Allergy and Clinical Immunology*, **2010**, 125, 170-4.e1-2	11.5	51
136	Raman profiles of the stratum corneum define 3 filaggrin genotype-determined atopic dermatitis endophenotypes. *Journal of Allergy and Clinical Immunology*, **2010**, 126, 574-80.e1	11.5	117
135	Chromosome 11q13.5 variant: No association with atopic eczema in the Japanese population. *Journal of Dermatological Science*, **2010**, 59, 210-2	4.3	1
134	Carrier status for the common R501X and 2282del4 filaggrin mutations is not associated with hearing phenotypes in 5,377 children from the ALSPAC cohort. *PLoS ONE*, **2009**, 4, e5784	3.7	7
133	Keratin 7 promoter selectively targets transgene expression to normal and neoplastic pancreatic ductal cells in vitro and in vivo. *FASEB Journal*, **2009**, 23, 1366-75	0.9	13
132	Identification of a novel family of laminin N-terminal alternate splice isoforms: structural and functional characterization. *Journal of Biological Chemistry*, **2009**, 284, 35588-96	5.4	20
131	Filaggrin loss-of-function variants are associated with atopic comorbidity in pediatric inflammatory bowel disease. *Inflammatory Bowel Diseases*, **2009**, 15, 1492-8	4.5	18
130	Ichthyosis vulgaris: novel FLG mutations in the German population and high presence of CD1a+ cells in the epidermis of the atopic subgroup. *British Journal of Dermatology*, **2009**, 160, 771-81	4	31
129	Analysis of Taiwanese ichthyosis vulgaris families further demonstrates differences in FLG mutations between European and Asian populations. *British Journal of Dermatology*, **2009**, 161, 448-51	4	39
128	Mutation in DSG1 causing autosomal dominant striate palmoplantar keratoderma. *British Journal of Dermatology*, **2009**, 161, 692-4	4	15
127	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, 884-9	4	77
126	Steatocystoma multiplex, oligodontia and partial persistent primary dentition associated with a novel keratin 17 mutation. *British Journal of Dermatology*, **2009**, 161, 1396-8	4	16
125	FLG mutation p.Lys4021X in the C-terminal imperfect filaggrin repeat in Japanese patients with atopic eczema. *British Journal of Dermatology*, **2009**, 161, 1387-90	4	59

124	Association of skin barrier genes within the PSORS4 locus is enriched in Singaporean Chinese with early-onset psoriasis. *Journal of Investigative Dermatology*, **2009**, 129, 606-14	4.3	22
123	Clinical severity correlates with impaired barrier in filaggrin-related eczema. *Journal of Investigative Dermatology*, **2009**, 129, 682-9	4.3	129
122	Heterozygous null alleles in filaggrin contribute to clinical dry skin in young adults and the elderly. *Journal of Investigative Dermatology*, **2009**, 129, 1042-5	4.3	47
121	Prevalent and rare mutations in the gene encoding filaggrin in Japanese patients with ichthyosis vulgaris and atopic dermatitis. *Journal of Investigative Dermatology*, **2009**, 129, 1302-5	4.3	35
120	Eczema genetics: current state of knowledge and future goals. *Journal of Investigative Dermatology* , **2009**, 129, 543-52	4.3	114
119	Achieving successful delivery of nucleic acids to skin: 6th Annual Meeting of the International Pachyonychia Congenita Consortium. *Journal of Investigative Dermatology*, **2009**, 129, 2085-7	4.3	13
118	Loss-of-function mutations of an inhibitory upstream ORF in the human hairless transcript cause Marie Unna hereditary hypotrichosis. *Nature Genetics*, **2009**, 41, 228-33	36.3	152
117	A homozygous frameshift mutation in the mouse Flg gene facilitates enhanced percutaneous allergen priming. *Nature Genetics*, **2009**, 41, 602-8	36.3	377
116	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, 1758-65	9.3	114
115	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, 807-13	11	53
114	Filaggrin in atopic dermatitis. *Journal of Allergy and Clinical Immunology*, **2009**, 124, R2-6	11.5	143
113	Filaggrin in the frontline: role in skin barrier function and disease. *Journal of Cell Science*, **2009**, 122, 1285-94	5.3	536
112	Mapping of two genetic loci for autosomal dominant hidradenitis suppurativa. *Experimental Dermatology*, **2008**, 15, 479-479	4	4
111	Unique and Recurrent Mutations in the Filaggrin Gene in Singaporean Chinese Patients with Ichthyosis Vulgaris. *Journal of Investigative Dermatology*, **2008**, 128, 1669-1675	4.3	51
110	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, 2117-9	4.3	228
109	Development of therapeutic siRNAs for pachyonychia congenita. *Journal of Investigative Dermatology*, **2008**, 128, 50-8	4.3	55
108	Single-nucleotide-specific siRNA targeting in a dominant-negative skin model. *Journal of Investigative Dermatology*, **2008**, 128, 594-605	4.3	90
107	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, 1323-5	4.3	35

106	Specific filaggrin mutations cause ichthyosis vulgaris and are significantly associated with atopic dermatitis in Japan. *Journal of Investigative Dermatology*, **2008**, 128, 1436-41	4.3	113
105	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, 1591-4	4.3	82
104	Filaggrin null mutations are associated with increased asthma exacerbations in children and young adults. *Allergy: European Journal of Allergy and Clinical Immunology*, **2008**, 63, 1211-7	9.3	39
103	Filaggrin null mutations and childhood atopic eczema: a population-based case-control study. *Journal of Allergy and Clinical Immunology*, **2008**, 121, 940-46.e3	11.5	121
102	The burden of disease associated with filaggrin mutations: a population-based, longitudinal birth cohort study. *Journal of Allergy and Clinical Immunology*, **2008**, 121, 872-7.e9	11.5	276
101	Filaggrin mutations, atopic eczema, hay fever, and asthma in children. *Journal of Allergy and Clinical Immunology*, **2008**, 121, 1203-1209.e1	11.5	317
100	Filaggrin variants confer susceptibility to asthma. *Journal of Allergy and Clinical Immunology*, **2008**, 121, 1294-5; author reply 1295-6	11.5	44
99	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, 560-8.e4	11.5	73
98	Filaggrin in atopic dermatitis. *Journal of Allergy and Clinical Immunology*, **2008**, 122, 689-693	11.5	267
97	Therapeutic siRNAs for dominant genetic skin disorders including pachyonychia congenita. *Journal of Dermatological Science*, **2008**, 51, 151-7	4.3	93
96	Broken bricks and cracked mortar: epidermal diseases resulting from genetic abnormalities. *Drug Discovery Today Disease Mechanisms*, **2008**, 5, e93-e101		6
95	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	11.6	187
94	The Human Intermediate Filament Database: comprehensive information on a gene family involved in many human diseases. *Human Mutation*, **2008**, 29, 351-60	4.7	270
93	Unique and recurrent mutations in the filaggrin gene in Singaporean Chinese patients with ichthyosis vulgaris. *Journal of Investigative Dermatology*, **2008**, 128, 1669-75	4.3	13
92	Unique mutations in the filaggrin gene in Japanese patients with ichthyosis vulgaris and atopic dermatitis. *Journal of Allergy and Clinical Immunology*, **2007**, 119, 434-40	11.5	191
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