W H Irwin Mclean

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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	Common loss-of-function variants of the epidermal barrier protein filaggrin are a major predisposing factor for atopic dermatitis. *Nature Genetics*, **2006**, 38, 441-6	36.3	2158
248	Loss-of-function mutations in the gene encoding filaggrin cause ichthyosis vulgaris. *Nature Genetics* , **2006**, 38, 337-42	36.3	804
247	Filaggrin mutations associated with skin and allergic diseases. *New England Journal of Medicine*, **2011**, 365, 1315-27	59.2	803
246	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
245	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nature Genetics*, **2012**, 44, 1341-8	36.3	681
244	Filaggrin in the frontline: role in skin barrier function and disease. *Journal of Cell Science*, **2009**, 122, 1285-94	5.3	536
243	Comprehensive analysis of the gene encoding filaggrin uncovers prevalent and rare mutations in ichthyosis vulgaris and atopic eczema. *Nature Genetics*, **2007**, 39, 650-4	36.3	510
242	Loss-of-function variations within the filaggrin gene predispose for atopic dermatitis with allergic sensitizations. *Journal of Allergy and Clinical Immunology*, **2006**, 118, 214-9	11.5	489
241	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
240	Intermediate filament proteins and their associated diseases. *New England Journal of Medicine*, **2004**, 351, 2087-100	59.2	392
239	A homozygous frameshift mutation in the mouse Flg gene facilitates enhanced percutaneous allergen priming. *Nature Genetics*, **2009**, 41, 602-8	36.3	377
238	Human keratin diseases: the increasing spectrum of disease and subtlety of the phenotype-genotype correlation. *British Journal of Dermatology*, **1999**, 140, 815-28	4	371
237	Plectin deficiency results in muscular dystrophy with epidermolysis bullosa. *Nature Genetics*, **1996**, 13, 450-7	36.3	358
236	One remarkable molecule: filaggrin. *Journal of Investigative Dermatology*, **2012**, 132, 751-62	4.3	343
235	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
234	Filaggrin mutations, atopic eczema, hay fever, and asthma in children. *Journal of Allergy and Clinical Immunology*, **2008**, 121, 1203-1209.e1	11.5	317
233	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

232	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
231	Filaggrin in atopic dermatitis. *Journal of Allergy and Clinical Immunology*, **2008**, 122, 689-693	11.5	267
230	Loss of kindlin-1, a human homolog of the Caenorhabditis elegans actin-extracellular-matrix linker protein UNC-112, causes Kindler syndrome. *American Journal of Human Genetics*, **2003**, 73, 174-87	11	263
229	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, 564-7	4.3	241
228	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
227	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. *Nature Genetics*, **2013**, 45, 1244-1248	36.3	217
226	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
225	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
224	Lipoid proteinosis maps to 1q21 and is caused by mutations in the extracellular matrix protein 1 gene (ECM1). *Human Molecular Genetics*, **2002**, 11, 833-40	5.6	192
223	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
222	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
221	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
220	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
219	Filaggrin genotype in ichthyosis vulgaris predicts abnormalities in epidermal structure and function. *American Journal of Pathology*, **2011**, 178, 2252-63	5.8	182
218	Mutations in cornea-specific keratin K3 or K12 genes cause Meesmann© corneal dystrophy. *Nature Genetics*, **1997**, 16, 184-7	36.3	181
217	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
216	Human keratin diseases: hereditary fragility of specific epithelial tissues. *Experimental Dermatology*, **1996**, 5, 297-307	4	180
215	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

214	Breaking the (un)sound barrier: filaggrin is a major gene for atopic dermatitis. *Journal of Investigative Dermatology*, **2006**, 126, 1200-2	4.3	177
213	Filaggrin null mutations are associated with increased asthma severity in children and young adults. *Journal of Allergy and Clinical Immunology*, **2007**, 120, 64-8	11.5	175
212	Prevalent and rare mutations in the gene encoding filaggrin cause ichthyosis vulgaris and predispose individuals to atopic dermatitis. *Journal of Investigative Dermatology*, **2006**, 126, 1770-5	4.3	173
211	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
210	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
209	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
208	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
207	Filaggrin in atopic dermatitis. *Journal of Allergy and Clinical Immunology*, **2009**, 124, R2-6	11.5	143
206	A mutation in human keratin K6b produces a phenocopy of the K17 disorder pachyonychia congenita type 2. *Human Molecular Genetics*, **1998**, 7, 1143-8	5.6	143
205	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
204	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
203	Clinical and pathological features of pachyonychia congenita. *Journal of Investigative Dermatology Symposium Proceedings*, **2005**, 10, 3-17	1.1	132
202	High-density genotyping study identifies four new susceptibility loci for atopic dermatitis. *Nature Genetics*, **2013**, 45, 808-12	36.3	131
201	Clinical severity correlates with impaired barrier in filaggrin-related eczema. *Journal of Investigative Dermatology*, **2009**, 129, 682-9	4.3	129
200	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, 1539-46	5.6	121
199	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
198	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
197	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

196	Eczema genetics: current state of knowledge and future goals. *Journal of Investigative Dermatology* , **2009**, 129, 543-52	4.3	114
195	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
194	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
193	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
192	Compound heterozygosity for non-sense and mis-sense mutations in desmoplakin underlies skin fragility/woolly hair syndrome. *Journal of Investigative Dermatology*, **2002**, 118, 232-8	4.3	109
191	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
190	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
189	A homozygous missense mutation in TGM5 abolishes epidermal transglutaminase 5 activity and causes acral peeling skin syndrome. *American Journal of Human Genetics*, **2005**, 77, 909-17	11	105
188	An unusual N-terminal deletion of the laminin alpha3a isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. *Human Molecular Genetics*, **2003**, 12, 2395-409	5.6	100
187	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
186	Role of transglutaminase 2 in glucose tolerance: knockout mice studies and a putative mutation in a MODY patient. *FASEB Journal*, **2002**, 16, 1371-8	0.9	99
185	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
184	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
183	Therapeutic siRNAs for dominant genetic skin disorders including pachyonychia congenita. *Journal of Dermatological Science*, **2008**, 51, 151-7	4.3	93
182	Plectin and human genetic disorders of the skin and muscle. The paradigm of epidermolysis bullosa with muscular dystrophy. *Experimental Dermatology*, **1996**, 5, 237-46	4	93
181	Single-nucleotide-specific siRNA targeting in a dominant-negative skin model. *Journal of Investigative Dermatology*, **2008**, 128, 594-605	4.3	90
180	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
179	Filaggrin© fuller figure: a glimpse into the genetic architecture of atopic dermatitis. *Journal of Investigative Dermatology*, **2007**, 127, 1282-4	4.3	86

178	Ichthyosis bullosa of Siemens--a disease involving keratin 2e. *Journal of Investigative Dermatology*, **1994**, 103, 277-81	4.3	86
177	The phenotypic and molecular genetic features of pachyonychia congenita. *Journal of Investigative Dermatology*, **2011**, 131, 1015-7	4.3	84
176	Ichthyosis bullosa of Siemens is caused by mutations in the keratin 2e gene. *Journal of Investigative Dermatology*, **1994**, 103, 286-9	4.3	84
175	Keratin disorders: from gene to therapy. *Human Molecular Genetics*, **2011**, 20, R189-97	5.6	83
174	Frameshift mutation in the V2 domain of human keratin 1 results in striate palmoplantar keratoderma. *Journal of Investigative Dermatology*, **2002**, 118, 838-44	4.3	83
173	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
172	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
171	Clinical and molecular characterization of lipoid proteinosis in Namaqualand, South Africa. *British Journal of Dermatology*, **2004**, 151, 413-23	4	79
170	The genetic basis of pachyonychia congenita. *Journal of Investigative Dermatology Symposium Proceedings*, **2005**, 10, 21-30	1.1	79
169	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
168	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
167	A novel connexin 30 mutation in Clouston syndrome. *Journal of Investigative Dermatology*, **2002**, 118, 530-2	4.3	71
166	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
165	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
164	The molecular genetics of the genodermatoses: progress to date and future directions. *British Journal of Dermatology*, **2003**, 148, 1-13	4	64
163	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
162	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	4.3	61
161	K15 expression implies lateral differentiation within stratified epithelial basal cells. *Laboratory Investigation*, **2000**, 80, 1701-10	5.9	61

160	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
159	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
158	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
157	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
156	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, 1391-6	4.3	57
155	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. *Nature Genetics*, **2012**, 44, 1272-6	36.3	56
154	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
153	Development of therapeutic siRNAs for pachyonychia congenita. *Journal of Investigative Dermatology*, **2008**, 128, 50-8	4.3	55
152	Clumping Factor B Promotes Adherence of Staphylococcus aureus to Corneocytes in Atopic Dermatitis. *Infection and Immunity*, **2017**, 85,	3.7	53
151	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
150	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
149	Epithelial inflammation resulting from an inherited loss-of-function mutation in EGFR. *Journal of Investigative Dermatology*, **2014**, 134, 2570-2578	4.3	51
148	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
147	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
146	Molecular genetics of Meesmann© corneal dystrophy: ancestral and novel mutations in keratin 12 (K12) and complete sequence of the human KRT12 gene. *Experimental Eye Research*, **2000**, 70, 41-9	3.7	50
145	Cyclic ichthyosis with epidermolytic hyperkeratosis: A phenotype conferred by mutations in the 2B domain of keratin K1. *American Journal of Human Genetics*, **1999**, 64, 732-8	11	50
144	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
143	Novel molecular therapies for heritable skin disorders. *Journal of Investigative Dermatology*, **2012**, 132, 820-8	4.3	49

142	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
141	Epidermolysis bullosa simplex in Israel: clinical and genetic features. *Archives of Dermatology*, **2003**, 139, 498-505		48
140	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
139	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
138	Novel mechanism of revertant mosaicism in Dowling-Meara epidermolysis bullosa simplex. *Journal of Investigative Dermatology*, **2004**, 122, 73-7	4.3	47
137	Statins downregulate K6a promoter activity: a possible therapeutic avenue for pachyonychia congenita. *Journal of Investigative Dermatology*, **2011**, 131, 1045-52	4.3	46
136	Epidermolysis bullosa: a spectrum of clinical phenotypes explained by molecular heterogeneity. *Trends in Molecular Medicine*, **1997**, 3, 457-65		46
135	A spectrum of mutations in keratins K6a, K16 and K17 causing pachyonychia congenita. *Journal of Dermatological Science*, **2007**, 48, 199-205	4.3	45
134	Novel keratin 16 mutations and protein expression studies in pachyonychia congenita type 1 and focal palmoplantar keratoderma. *Experimental Dermatology*, **2000**, 9, 170-7	4	45
133	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, 413-9	11	45
132	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
131	Filaggrin variants confer susceptibility to asthma. *Journal of Allergy and Clinical Immunology*, **2008**, 121, 1294-5; author reply 1295-6	11.5	44
130	Filaggrin mutations are genetic modifying factors exacerbating X-linked ichthyosis. *Journal of Investigative Dermatology*, **2007**, 127, 2795-8	4.3	44
129	Defolliculated (dfl): a dominant mouse mutation leading to poor sebaceous gland differentiation and total elimination of pelage follicles. *Journal of Investigative Dermatology*, **2002**, 119, 32-7	4.3	44
128	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, 687-91	4.9	44
127	A large mutational study in pachyonychia congenita. *Journal of Investigative Dermatology*, **2011**, 131, 1018-24	4.3	43
126	cDNA cloning, mRNA expression, and chromosomal mapping of human and mouse periplakin genes. *Genomics*, **1998**, 48, 242-7	4.3	43
125	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

124	Clouston syndrome can mimic pachyonychia congenita. *Journal of Investigative Dermatology*, **2003**, 121, 1035-8	4.3	42
123	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
122	Two cases of primarily palmoplantar keratoderma associated with novel mutations in keratin 1. *Journal of Investigative Dermatology*, **2002**, 119, 966-71	4.3	40
121	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
120	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
119	A heterozygous frameshift mutation in the V1 domain of keratin 5 in a family with Dowling-Degos disease. *Journal of Investigative Dermatology*, **2007**, 127, 298-300	4.3	39
118	Atypical epidermolytic palmoplantar keratoderma presentation associated with a mutation in the keratin 1 gene. *British Journal of Dermatology*, **2004**, 150, 1096-103	4	39
117	Mutations in keratin K9 in kindreds with epidermolytic palmoplantar keratoderma and epidemiology in Northern Ireland. *Journal of Investigative Dermatology*, **1998**, 111, 1207-9	4.3	38
116	Delayed-onset pachyonychia congenita associated with a novel mutation in the central 2B domain of keratin 16. *British Journal of Dermatology*, **2001**, 144, 1058-62	4	38
115	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
114	Mutations in GRHL2 result in an autosomal-recessive ectodermal Dysplasia syndrome. *American Journal of Human Genetics*, **2014**, 95, 308-14	11	37
113	Keratin K6c mutations cause focal palmoplantar keratoderma. *Journal of Investigative Dermatology*, **2010**, 130, 425-9	4.3	37
112	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex. *Human Mutation*, **1998**, 11, 279-85	4.7	36
111	Filaggrin null alleles are not associated with psoriasis. *Journal of Investigative Dermatology*, **2007**, 127, 1878-82	4.3	36
110	A mutation in the V1 domain of K16 is responsible for unilateral palmoplantar verrucous nevus. *Journal of Investigative Dermatology*, **2000**, 114, 1136-40	4.3	36
109	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
108	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
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	Insights into genotype-phenotype correlation in pachyonychia congenita from the human intermediate filament mutation database. *Journal of Investigative Dermatology Symposium Proceedings*, **2005**, 10, 31-6	1.1	35
105	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
104	Generic and personalized RNAi-based therapeutics for a dominant-negative epidermal fragility disorder. *Journal of Investigative Dermatology*, **2012**, 132, 1627-35	4.3	34
103	Focal palmoplantar keratoderma caused by an autosomal dominant inherited mutation in the desmoglein 1 gene. *Dermatology*, **2006**, 212, 117-22	4.4	33
102	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
101	Keratin K6irs is specific to the inner root sheath of hair follicles in mice and humans. *British Journal of Dermatology*, **2001**, 145, 558-68	4	32
100	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
99	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
98	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
97	Development of allele-specific therapeutic siRNA in Meesmann epithelial corneal dystrophy. *PLoS ONE*, **2011**, 6, e28582	3.7	30
96	Allele-specific siRNA silencing for the common keratin 12 founder mutation in Meesmann epithelial corneal dystrophy **2013**, 54, 494-502		29
95	Genomic organization and fine mapping of the keratin 2e gene (KRT2E): K2e V1 domain polymorphism and novel mutations in ichthyosis bullosa of Siemens. *Journal of Investigative Dermatology*, **1998**, 111, 817-21	4.3	28
94	Mutation S233L in the 1B domain of keratin 1 causes epidermolytic palmoplantar keratoderma with "tonotubular" keratin. *Journal of Investigative Dermatology*, **2006**, 126, 607-13	4.3	28
93	Genetic disorders of palm skin and nail. *Journal of Anatomy*, **2003**, 202, 133-41	2.9	28
92	Cloning of multiple keratin 16 genes facilitates prenatal diagnosis of pachyonychia congenita type 1 **1999**, 19, 941-946		28
91	A novel mutation in KRT12 associated with Meesmann© epithelial corneal dystrophy. *British Journal of Ophthalmology*, **2002**, 86, 729-32	5.5	27
90	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
89	Plectin mutations underlie epidermolysis bullosa simplex in 8% of patients. *Journal of Investigative Dermatology*, **2014**, 134, 273-276	4.3	26

88	Kindler surprise: mutations in a novel actin-associated protein cause Kindler syndrome. *Journal of Dermatological Science*, **2005**, 38, 169-75	4.3	26
87	Cloning of human, murine, and marsupial keratin 7 and a survey of K7 expression in the mouse. *Biochemical and Biophysical Research Communications*, **2002**, 297, 818-27	3.4	26
86	Breach delivery: increased solute uptake points to a defective skin barrier in atopic dermatitis. *Journal of Investigative Dermatology*, **2007**, 127, 8-10	4.3	25
85	Novel proline substitution mutations in keratin 16 in two cases of pachyonychia congenita type 1. *British Journal of Dermatology*, **1999**, 141, 1010-6	4	25
84	Generation and characterisation of keratin 7 (K7) knockout mice. *PLoS ONE*, **2013**, 8, e64404	3.7	24
83	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
82	A novel keratin 12 mutation in a German kindred with Meesmann corneal dystrophy. *British Journal of Ophthalmology*, **2000**, 84, 527-30	5.5	24
81	A new, recurrent mutation of GJB3 (Cx31) in erythrokeratodermia variabilis. *British Journal of Dermatology*, **2005**, 152, 1143-8	4	23
80	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. *American Journal of Human Genetics*, **2017**, 100, 364-370	11	22
79	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
78	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
77	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
76	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
75	An atypical form of bullous congenital ichthyosiform erythroderma is caused by a mutation in the L12 linker region of keratin 1. *Journal of Investigative Dermatology*, **1998**, 111, 1224-6	4.3	21
74	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
73	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
72	Expanding the Phenotypic Spectrum of Olmsted Syndrome. *Journal of Investigative Dermatology*, **2015**, 135, 2879-2883	4.3	19
71	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

70	Functional analysis of keratin components in the mouse hair follicle inner root sheath. *British Journal of Dermatology*, **2004**, 150, 195-204	4	19
69	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
68	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
67	Heritable filaggrin disorders: the paradigm of atopic dermatitis. *Journal of Investigative Dermatology*, **2012**, 132, E20-1	4.3	18
66	Long-range polymerase chain reaction for specific full-length amplification of the human keratin 14 gene and novel keratin 14 mutations in epidermolysis bullosa simplex patients. *Journal of Investigative Dermatology*, **2003**, 120, 495-7	4.3	18
65	The gene for Naegeli-Franceschetti-Jadassohn syndrome maps to 17q21. *Journal of Investigative Dermatology*, **2000**, 115, 694-8	4.3	18
64	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
63	Gentamicin-Induced Readthrough and Nonsense-Mediated mRNA Decay of SERPINB7 Nonsense Mutant Transcripts. *Journal of Investigative Dermatology*, **2018**, 138, 836-843	4.3	17
62	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
61	Development of quantitative molecular clinical end points for siRNA clinical trials. *Journal of Investigative Dermatology*, **2011**, 131, 1029-36	4.3	17
60	SiRNA-mediated selective inhibition of mutant keratin mRNAs responsible for the skin disorder pachyonychia congenita. *Annals of the New York Academy of Sciences*, **2006**, 1082, 56-61	6.5	17
59	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
58	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
57	Mutation in DSG1 causing autosomal dominant striate palmoplantar keratoderma. *British Journal of Dermatology*, **2009**, 161, 692-4	4	15
56	Follow up of patients with ocular scarring secondary to LOC syndrome treated by amniotic membrane transplantation. *British Journal of Ophthalmology*, **2005**, 89, 939-41	5.5	15
55	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
54	Human periplakin: genomic organization in a clonally unstable region of chromosome 16p with an abundance of repetitive sequence elements. *Genomics*, **1999**, 56, 160-8	4.3	14
53	Disorders of keratinisation: from rare to common genetic diseases of skin and other epithelial tissues. *Ulster Medical Journal*, **2007**, 76, 72-82	0.4	14

52	Discovery of super soft-drug modulators of sphingosine-1-phosphate receptor 1. *Bioorganic and Medicinal Chemistry Letters*, **2018**, 28, 3255-3259	2.9	13
51	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
50	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
49	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
48	Rapid mutation detection by the transgenomic wave analyser DHPLC identifies MYOC mutations in patients with ocular hypertension and/or open angle glaucoma. *British Journal of Ophthalmology*, **2002**, 86, 191-5	5.5	13
47	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
46	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
45	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
44	Characterization of human skin fibroblast extracellular proteins by two-dimensional polyacrylamide gel electrophoresis. *Electrophoresis*, **1988**, 9, 343-51	3.6	11
43	The allergy gene: how a mutation in a skin protein revealed a link between eczema and asthma. *F1000 Medicine Reports*, **2011**, 3, 2		11
42	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
41	Novel TGM5 mutations in acral peeling skin syndrome. *Experimental Dermatology*, **2015**, 24, 285-9	4	10
40	Possibilities for human skin characterization based on strongly reduced Raman spectroscopic information. *Journal of Raman Spectroscopy*, **2013**, 44, 340-345	2.3	10
39	Gut microbiota development during infancy: Impact of introducing allergenic foods. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 613-621.e9	11.5	10
38	Filaggrin gene loss-of-function mutations constitute a factor in patients with multiple contact allergies. *Contact Dermatitis*, **2019**, 80, 354-358	2.7	9
37	Genomic localization, organization and amplification of the human zinc transporter protein gene, ZNT4, and exclusion as a candidate gene in different clinical variants of acrodermatitis enteropathica. *Archives of Dermatological Research*, **2001**, 293, 392-6	3.3	9
36	Mapping of the human CP49 gene and identification of an intragenic polymorphic marker to allow genetic linkage analysis in autosomal dominant congenital cataract. *Biochemical and Biophysical Research Communications*, **2000**, 270, 432-6	3.4	9
35	Punctate palmoplantar keratoderma type 1: a novel AAGAB mutation and efficacy of etretinate. *Acta Dermato-Venereologica*, **2015**, 95, 110-1	2.2	8

34	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
33	Reliability and validity of genotyping filaggrin null mutations. *Journal of Dermatological Science*, **2013**, 70, 67-8	4.3	8
32	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
31	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
30	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
29	Close shave for a keratin disorder-K6hf polymorphism linked to Pseudofolliculitis barbae. *Journal of Investigative Dermatology*, **2004**, 122, xi-xiii	4.3	8
28	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
27	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
26	Genomic organization, amplification, fine mapping, and intragenic polymorphisms of the human hemidesmosomal tetraspanin CD151 gene. *Biochemical and Biophysical Research Communications*, **2001**, 281, 425-30	3.4	7
25	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
24	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
23	Broken bricks and cracked mortar epidermal diseases resulting from genetic abnormalities. *Drug Discovery Today Disease Mechanisms*, **2008**, 5, e93-e101		6
22	A novel mutation in the 2B domain of keratin 2e causing ichthyosis bullosa of Siemens. *Clinical and Experimental Dermatology*, **2000**, 25, 648-51	1.8	6
21	Old King coal - molecular mechanisms underlying an ancient treatment for atopic eczema. *Journal of Clinical Investigation*, **2013**, 123, 551-3	15.9	6
20	Paternal germ cell mosaicism in autosomal dominant pachyonychia congenita. *Archives of Dermatology*, **2011**, 147, 1077-80		4
19	Mapping of two genetic loci for autosomal dominant hidradenitis suppurativa. *Experimental Dermatology*, **2008**, 15, 479-479	4	4
18	Molecular genetics methods for human intermediate filament diseases. *Methods in Cell Biology*, **2004**, 78, 131-61	1.8	4
17	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

16	PCQoL: A Quality of Life Assessment Measure for Pachyonychia Congenita. *Journal of Cutaneous Medicine and Surgery*, **2015**, 19, 57-65	1.6	3
15	Transgrediens pachyonychia congenita (PC): case series of a nonclassical PC presentation. *British Journal of Dermatology*, **2012**, 166, 124-8	4	3
14	Resolution of the plantar hyperkeratosis of pachyonychia congenita during chemotherapy for Ewing sarcoma. *British Journal of Dermatology*, **2013**, 169, 1357-60	4	3
13	AvaII RFLP of human keratin 10 (KRT-10) detected by PCR. *Human Molecular Genetics*, **1992**, 1, 659	5.6	3
12	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
11	Male genital lichen sclerosus and filaggrin. *Clinical and Experimental Dermatology*, **2020**, 45, 127-128	1.8	3
10	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex **1998**, 11, 279		3
9	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
8	A new filaggrin gene mutation in a Korean patient with ichthyosis vulgaris. *European Journal of Dermatology*, **2014**, 24, 491-3	0.8	2
7	Genetics of Hidradenitis Suppurativa **2006**, 70-85		2
6	Cloning of multiple keratin 16 genes facilitates prenatal diagnosis of pachyonychia congenita type 1 **1999**, 19, 941		2
5	Genodermatoses: Inherited Diseases of the Skin **2011**, 379-409		1
4	Chromosome 11q13.5 variant: No association with atopic eczema in the Japanese population. *Journal of Dermatological Science*, **2010**, 59, 210-2	4.3	1
3	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
2	Localization of a human fibroblast extracellular protein in cells and tissues by monoclonal antibody. *Biochemical Society Transactions*, **1990**, 18, 279-80	5.1	
1	Reduced secretion of specific proteins in dystrophic fibroblasts. *Biochemical Society Transactions*, **1990**, 18, 281-2	5.1	