

J E Mellerio

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

140
papers

5,613
citations

39
h-index

71
g-index

162
ext. papers

6,585
ext. citations

2.7
avg, IF

5.25
L-index

#	Paper	IF	Citations
140	Characteristics of children with Netherton syndrome: a review of 21 patients. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2021 , 35, e466-e469	4.6	1
139	Heterogeneous addiction to transforming growth factor-beta signalling in recessive dystrophic epidermolysis bullosa-associated cutaneous squamous cell carcinoma. <i>British Journal of Dermatology</i> , 2021 , 184, 697-708	4	3
138	Prevalence, pathophysiology and management of itch in epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2021 , 184, 816-825	4	12
137	Practical management of epidermolysis bullosa: consensus clinical position statement from the European Reference Network for Rare Skin Diseases. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2021 , 35, 2349-2360	4.6	2
136	The epidemiology of epidermolysis bullosa in England and Wales: data from the national epidermolysis bullosa database.. <i>British Journal of Dermatology</i> , 2021 ,	4	1
135	Meeting Report: The First Global Congress on Epidermolysis Bullosa, EB2020 London: Toward Treatment and Cure. <i>Journal of Investigative Dermatology</i> , 2020 , 140, 1681-1687	4.3	8
134	EBGene trial: patient preselection outcomes for the European GENEGRAFT ex vivo phase I/II gene therapy trial for recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2020 , 182, 794-797	4	15
133	The psychological functioning of children with epidermolysis bullosa and its relationship with specific aspects of disease. <i>British Journal of Dermatology</i> , 2020 , 182, 789-790	4	2
132	Pseudoporphyria induced by ultraviolet radiation. <i>Australasian Journal of Dermatology</i> , 2020 , 61, 177-179.	3	3
131	Homozygous Nonsense Mutation in DSC3 Resulting in Skin Fragility and Hypotrichosis. <i>Journal of Investigative Dermatology</i> , 2020 , 140, 1285-1288	4.3	4
130	Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. <i>British Journal of Dermatology</i> , 2020 , 183, 614-627	4	179
129	PLACK syndrome: the penny dropped. <i>Clinical and Experimental Dermatology</i> , 2020 , 45, 1091-1092	1.8	0
128	Genotype-phenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. <i>British Journal of Dermatology</i> , 2020 , 182, 729-737	4	19
127	Foot care in epidermolysis bullosa: evidence-based guideline. <i>British Journal of Dermatology</i> , 2020 , 182, 593-604	4	9
126	Beta blockers for infantile haemangiomas: where should we go from here?. <i>British Journal of Dermatology</i> , 2019 , 180, 450-451	4	0
125	Potential therapeutic targeting of inflammation in epidermolysis bullosa simplex. <i>British Journal of Dermatology</i> , 2019 , 180, 258-260	4	3
124	Itch and scratch: could pain be the catch?. <i>British Journal of Dermatology</i> , 2018 , 179, 1029	4	0

123	Predictive phenotyping of inherited ichthyosis by next-generation DNA sequencing. <i>British Journal of Dermatology</i> , 2017 , 176, 249-251	4	6
122	Focal dermal hypoplasia: inheritance from father to daughter. <i>Clinical and Experimental Dermatology</i> , 2017 , 42, 457-459	1.8	3
121	Wound healing in epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2017 , 177, e193-e195	4	6
120	Neonatal aggressive systemic mastocytosis. <i>British Journal of Dermatology</i> , 2017 , 177, 1167-1168	4	
119	Considerations in surgical management of a Buschke-Lowenstein tumor in Netherton syndrome: A case report. <i>Pediatric Dermatology</i> , 2017 , 34, e328-e330	1.9	7
118	Ichthyosis Prematurity Syndrome: From Fetus to Adulthood. <i>JAMA Dermatology</i> , 2016 , 152, 1055-8	5.1	6
117	Management of cutaneous squamous cell carcinoma in patients with epidermolysis bullosa: best clinical practice guidelines. <i>British Journal of Dermatology</i> , 2016 , 174, 56-67	4	70
116	Autosomal dominant diffuse nonepidermolytic palmoplantar keratoderma due to a recurrent mutation in aquaporin-5. <i>British Journal of Dermatology</i> , 2016 , 174, 430-2	4	7
115	Mutations in EXPH5 underlie a rare subtype of autosomal recessive epidermolysis bullosa simplex. <i>British Journal of Dermatology</i> , 2016 , 174, 452-3	4	7
114	Early-onset dermatosis papulosa nigra. <i>British Journal of Dermatology</i> , 2016 , 174, 1148-50	4	6
113	Potential of Systemic Allogeneic Mesenchymal Stromal Cell Therapy for Children with Recessive Dystrophic Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , 2015 , 135, 2319-2321	4.3	89
112	Pityriasis rubra pilaris with histologic features of lichen nitidus. <i>Journal of the American Academy of Dermatology</i> , 2015 , 73, 336-7	4.5	3
111	Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. <i>British Journal of Dermatology</i> , 2015 , 172, 94-100	4	54
110	Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. <i>Journal of the American Academy of Dermatology</i> , 2014 , 70, 1103-26	4.5	596
109	Under-recognition of acral peeling skin syndrome: 59 new cases with 15 novel mutations. <i>British Journal of Dermatology</i> , 2014 , 171, 1206-10	4	21
108	Epithelial inflammation resulting from an inherited loss-of-function mutation in EGFR. <i>Journal of Investigative Dermatology</i> , 2014 , 134, 2570-2578	4.3	51
107	Clinical features and WNT10A mutations in seven unrelated cases of Schöpf-Schulz-Passarge syndrome. <i>British Journal of Dermatology</i> , 2014 , 171, 1211-4	4	20
106	The missense mutation p.R1303Q in type XVII collagen underlies junctional epidermolysis bullosa resembling Kindler syndrome. <i>Journal of Investigative Dermatology</i> , 2014 , 134, 845-849	4.3	16

105	G45 Haematocolpos due to a transverse vaginal septum in 15 year old girl with junctional epidermolysis bullosa. <i>Archives of Disease in Childhood</i> , 2014 , 99, A19-A19	2.2	1
104	Mutations in EXPH5 result in autosomal recessive inherited skin fragility. <i>British Journal of Dermatology</i> , 2014 , 170, 196-9	4	9
103	Pain, purpura and curly hairs. <i>Clinical and Experimental Dermatology</i> , 2013 , 38, 940-2	1.8	6
102	Serum levels of high mobility group box 1 correlate with disease severity in recessive dystrophic epidermolysis bullosa. <i>Experimental Dermatology</i> , 2013 , 22, 433-5	4	22
101	Fibroblast cell therapy enhances initial healing in recessive dystrophic epidermolysis bullosa wounds: results of a randomized, vehicle-controlled trial. <i>British Journal of Dermatology</i> , 2013 , 169, 1025-33	4.33	9 ²
100	Phase I study protocol for ex vivo lentiviral gene therapy for the inherited skin disease, Netherton syndrome. <i>Human Gene Therapy Clinical Development</i> , 2013 , 24, 182-90	3.2	30
99	Homozygous variegate porphyria presenting with developmental and language delay in childhood. <i>Clinical and Experimental Dermatology</i> , 2013 , 38, 737-40	1.8	11
98	Mutations in AEC syndrome skin reveal a role for p63 in basement membrane adhesion, skin barrier integrity and hair follicle biology. <i>British Journal of Dermatology</i> , 2012 , 167, 134-44	4	22
97	MBTPS2 mutation in a British pedigree with keratosis follicularis spinulosa decalvans. <i>Clinical and Experimental Dermatology</i> , 2012 , 37, 631-4	1.8	12
96	A consensus approach to wound care in epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , 2012 , 67, 904-17	4.5	103
95	Gastrostomy tube feeding in children with epidermolysis bullosa: consideration of key issues. <i>Pediatric Dermatology</i> , 2012 , 29, 277-84	1.9	16
94	A pyrexial unwell child with a papular eruption. <i>Clinical and Experimental Dermatology</i> , 2012 , 37, 811-3	1.8	2
93	Novel and recurrent COL7A1 mutations in Chilean patients with dystrophic epidermolysis bullosa. <i>Journal of Dermatological Science</i> , 2012 , 65, 149-52	4.3	14
92	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. <i>American Journal of Human Genetics</i> , 2012 , 91, 1115-21	11	48
91	Desmosomal genodermatoses. <i>British Journal of Dermatology</i> , 2012 , 166, 36-45	4	59
90	Immunofluorescence antigen mapping for hereditary epidermolysis bullosa. <i>Indian Journal of Dermatology, Venereology and Leprology</i> , 2012 , 78, 692-7	0.8	17
89	Autosomal recessive epidermolysis bullosa simplex due to loss of BPAG1-e expression. <i>Journal of Investigative Dermatology</i> , 2012 , 132, 742-4	4.3	39
88	Fluoroscopically guided dilation of esophageal strictures in patients with dystrophic epidermolysis bullosa: long-term results. <i>American Journal of Roentgenology</i> , 2012 , 199, 208-12	5.4	26

87	Growth Impaired Children with Epidermolysis Bullosa have Increased Serum Markers of Inflammation and Reduced Circulating IGF-1/IGFBP-3. <i>Pediatric Research</i> , 2011 , 70, 294-294	3.2	2
86	Recalcitrant generalized eruption and low alkaline phosphatase: think zinc. <i>Clinical and Experimental Dermatology</i> , 2011 , 36, 225-6	1.8	1
85	The challenges of meeting nutritional requirements in children and adults with epidermolysis bullosa: proceedings of a multidisciplinary team study day. <i>Clinical and Experimental Dermatology</i> , 2011 , 36, 579-83; quiz 583-4	1.8	18
84	An unusual case of epidermolysis bullosa complicated by persistent oligoarticular juvenile idiopathic arthritis; lessons to be learned. <i>Pediatric Rheumatology</i> , 2011 , 9, 13	3.5	2
83	The international dystrophic epidermolysis bullosa patient registry: an online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. <i>Human Mutation</i> , 2011 , 32, 1100-7	4.7	63
82	The inversa type of recessive dystrophic epidermolysis bullosa is caused by specific arginine and glycine substitutions in type VII collagen. <i>Journal of Medical Genetics</i> , 2011 , 48, 160-7	5.8	21
81	Harlequin ichthyosis: a review of clinical and molecular findings in 45 cases. <i>Archives of Dermatology</i> , 2011 , 147, 681-6		111
80	HB-EGF induces COL7A1 expression in keratinocytes and fibroblasts: possible mechanism underlying allogeneic fibroblast therapy in recessive dystrophic epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , 2011 , 131, 1771-4	4.3	54
79	Identical glycine substitution mutations in type VII collagen may underlie both dominant and recessive forms of dystrophic epidermolysis bullosa. <i>Acta Dermato-Venereologica</i> , 2011 , 91, 262-6	2.2	28
78	Molecular basis of EEC (ectrodactyly, ectodermal dysplasia, clefting) syndrome: five new mutations in the DNA-binding domain of the TP63 gene and genotype-phenotype correlation. <i>British Journal of Dermatology</i> , 2010 , 162, 201-7	4	31
77	Rapp-Hodgkin and Hay-Wells ectodermal dysplasia syndromes represent a variable spectrum of the same genetic disorder. <i>British Journal of Dermatology</i> , 2010 , 163, 624-9	4	29
76	Bullous pemphigoid in a patient with suspected non-Herlitz junctional epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 2010 , 35, 881-4	1.8	2
75	Revertant mosaicism in recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , 2010 , 130, 1937-40	4.3	47
74	A homozygous nonsense mutation within the dystonin gene coding for the coiled-coil domain of the epithelial isoform of BPAG1 underlies a new subtype of autosomal recessive epidermolysis bullosa simplex. <i>Journal of Investigative Dermatology</i> , 2010 , 130, 1551-7	4.3	101
73	Homozygous mutations in the 5' region of the JUP gene result in cutaneous disease but normal heart development in children. <i>Journal of Investigative Dermatology</i> , 2010 , 130, 1543-50	4.3	40
72	Ectodermal dysplasia-skin fragility syndrome. <i>Dermatologic Clinics</i> , 2010 , 28, 125-9	4.2	55
71	Epidermolysis bullosa care in the United Kingdom. <i>Dermatologic Clinics</i> , 2010 , 28, 395-6, xiv	4.2	6
70	Infection and colonization in epidermolysis bullosa. <i>Dermatologic Clinics</i> , 2010 , 28, 267-9, ix	4.2	40

69	Congenital muscular dystrophy, myasthenic symptoms and epidermolysis bullosa simplex (EBS) associated with mutations in the PLEC1 gene encoding plectin. <i>Neuromuscular Disorders</i> , 2010 , 20, 709-11	9	35
68	Osteopenia and osteoporosis in epidermolysis bullosa. <i>Dermatologic Clinics</i> , 2010 , 28, 353-5, xi	4.2	16
67	Genitourinary tract involvement in epidermolysis bullosa. <i>Dermatologic Clinics</i> , 2010 , 28, 343-6, xi	4.2	9
66	Genetics and Genodermatoses 2010 , 1-97		12
65	Dilated cardiomyopathy in epidermolysis bullosa: a retrospective, multicenter study. <i>Pediatric Dermatology</i> , 2010 , 27, 238-43	1.9	27
64	New glycine substitution mutations in type VII collagen underlying epidermolysis bullosa pruriginosa but the phenotype is not explained by a common polymorphism in the matrix metalloproteinase-1 gene promoter. <i>Acta Dermato-Venereologica</i> , 2009 , 89, 6-11	2.2	29
63	Autosomal dominant junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2009 , 160, 1094-7	4	28
62	PORCN gene mutations and the protean nature of focal dermal hypoplasia. <i>British Journal of Dermatology</i> , 2009 , 160, 1103-9	4	32
61	Epidermolysis bullosa pruriginosa in association with lichen planopilaris. <i>Clinical and Experimental Dermatology</i> , 2009 , 34, e825-8	1.8	10
60	Maxillary alveolar process fracture complicating intubation in a patient with epidermolysis bullosa. <i>Paediatric Anaesthesia</i> , 2009 , 19, 706-7	1.8	3
59	Extracutaneous manifestations and complications of inherited epidermolysis bullosa: part I. Epithelial associated tissues. <i>Journal of the American Academy of Dermatology</i> , 2009 , 61, 367-84; quiz 385-6	4.5	165
58	Extracutaneous manifestations and complications of inherited epidermolysis bullosa: part II. Other organs. <i>Journal of the American Academy of Dermatology</i> , 2009 , 61, 387-402; quiz 403-4	4.5	155
57	Potential of fibroblast cell therapy for recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , 2008 , 128, 2179-89	4.3	176
56	Congenital anetoderma in a preterm infant. <i>Pediatric Dermatology</i> , 2008 , 25, 626-9	1.9	10
55	Gastrointestinal complications of epidermolysis bullosa in children. <i>British Journal of Dermatology</i> , 2008 , 158, 1308-14	4	80
54	Anaesthetic management of two different modes of delivery in patients with dystrophic epidermolysis bullosa. <i>International Journal of Obstetric Anesthesia</i> , 2008 , 17, 153-8	2.1	18
53	The classification of inherited epidermolysis bullosa (EB): Report of the Third International Consensus Meeting on Diagnosis and Classification of EB. <i>Journal of the American Academy of Dermatology</i> , 2008 , 58, 931-50	4.5	690
52	Novel and recurring ABCA12 mutations associated with harlequin ichthyosis: implications for prenatal diagnosis. <i>British Journal of Dermatology</i> , 2008 , 158, 611-3	4	21

51	Generalized pustular eruption in a 5-year-old boy. <i>Clinical and Experimental Dermatology</i> , 2008 , 33, 79-80.	1.8	1
50	A spectrum of mutations in keratins K6a, K16 and K17 causing pachyonychia congenita. <i>Journal of Dermatological Science</i> , 2007 , 48, 199-205	4.3	45
49	Plectin defects in epidermolysis bullosa simplex with muscular dystrophy. <i>Muscle and Nerve</i> , 2007 , 35, 24-35	3.4	54
48	A heterozygous frameshift mutation in the V1 domain of keratin 5 in a family with Dowling-Degos disease. <i>Journal of Investigative Dermatology</i> , 2007 , 127, 298-300	4.3	39
47	Histopathological features of gastrointestinal mucosal biopsy specimens in children with epidermolysis bullosa. <i>Journal of Clinical Pathology</i> , 2007 , 60, 843-4	3.9	6
46	The management of general and disease specific ENT problems in children with Epidermolysis Bullosa--a retrospective case note review. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2007 , 71, 385-91	1.7	21
45	Epidermolysis bullosa. <i>British Journal of Hospital Medicine (London, England: 2005)</i> , 2006 , 67, 188-91	0.8	16
44	Prenatal diagnosis for severe inherited skin disorders: 25 years experience. <i>British Journal of Dermatology</i> , 2006 , 154, 106-13	4	64
43	Bone mineralization in children with epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2006 , 154, 959-62	4	46
42	Target proteins in inherited and acquired blistering skin disorders. <i>Clinical and Experimental Dermatology</i> , 2006 , 31, 252-9	1.8	30
41	An unusual patient with Rothmund-Thomson syndrome, porokeratosis and bilateral iris dysgenesis. <i>Clinical and Experimental Dermatology</i> , 2006 , 31, 401-3	1.8	14
40	Complete maternal isodisomy of chromosome 3 in a child with recessive dystrophic epidermolysis bullosa but no other phenotypic abnormalities. <i>Journal of Investigative Dermatology</i> , 2006 , 126, 2039-43	4.3	28
39	Neonatal diagnosis of Kindler syndrome. <i>Journal of Dermatological Science</i> , 2005 , 39, 183-5	4.3	19
38	Thalidomide in the management of epidermolysis bullosa pruriginosa. <i>British Journal of Dermatology</i> , 2005 , 152, 1332-4	4	44
37	Complete paternal uniparental isodisomy of chromosome 1 resulting in Herlitz junctional epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 2005 , 30, 71-4	1.8	26
36	Multiple dermatofibromas associated with lupus profundus. <i>Clinical and Experimental Dermatology</i> , 2005 , 30, 128-30	1.8	10
35	Prenatal diagnosis of Herlitz junctional epidermolysis bullosa in nonidentical twins. <i>Clinical and Experimental Dermatology</i> , 2005 , 30, 180-2	1.8	5
34	ADULT ectodermal dysplasia syndrome resulting from the missense mutation R298Q in the p63 gene. <i>Clinical and Experimental Dermatology</i> , 2004 , 29, 669-72	1.8	26

33	An unusual N-terminal deletion of the laminin alpha3a isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. <i>Human Molecular Genetics</i> , 2003 , 12, 2395-409	5.6	100
32	Dilemmas in distinguishing between dominant and recessive forms of dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2003 , 149, 810-8	4	25
31	Skin disease in Gulf war veterans. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2002 , 95, 671-6	2.7	4
30	Lipoid proteinosis maps to 1q21 and is caused by mutations in the extracellular matrix protein 1 gene (ECM1). <i>Human Molecular Genetics</i> , 2002 , 11, 833-40	5.6	192
29	Alpha 6 beta 4 integrin abnormalities in junctional epidermolysis bullosa with pyloric atresia. <i>British Journal of Dermatology</i> , 2001 , 144, 408-14	4	52
28	Three cases of de novo dominant dystrophic epidermolysis bullosa associated with the mutation G2043R in COL7A1. <i>Clinical and Experimental Dermatology</i> , 2001 , 26, 97-9	1.8	14
27	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , 2001 , 26, 93-96	1.8	3
26	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , 2001 , 26, 93-96	1.8	1
25	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , 2001 , 26, 93-6	1.8	20
24	Heterogeneous Mutations of the Type VII Collagen Gene (COL7A1) in Dystrophic Epidermolysis Bullosa. <i>Clinical Science</i> , 2000 , 98, 18P-18P		
23	Heterozygous germline missense mutation in the p63 gene underlying EEC syndrome. <i>Clinical and Experimental Dermatology</i> , 2000 , 25, 441-3	1.8	33
22	Moderation of phenotypic severity in dystrophic and junctional forms of epidermolysis bullosa through in-frame skipping of exons containing non-sense or frameshift mutations. <i>Journal of Investigative Dermatology</i> , 1999 , 113, 314-21	4.3	53
21	Skin fragility and hypohidrotic ectodermal dysplasia resulting from ablation of plakophilin 1. <i>British Journal of Dermatology</i> , 1999 , 140, 297-307	4	92
20	A recurrent COL7A1 mutation, R2814X, in British patients with recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 1999 , 24, 37-9	1.8	10
19	Frequency of the CCR5 gene 32-basepair deletion in Hispanic Mexicans. <i>Clinical and Experimental Dermatology</i> , 1999 , 24, 127-9	1.8	5
18	Recurrent molecular abnormalities in type VII collagen in Southern Italian patients with recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 1999 , 24, 232-5	1.8	8
17	Allelic heterogeneity of dominant and recessive COL7A1 mutations underlying epidermolysis bullosa pruriginosa. <i>Journal of Investigative Dermatology</i> , 1999 , 112, 984-7	4.3	59
16	Comparative mutation detection screening of the type VII collagen gene (COL7A1) using the protein truncation test, fluorescent chemical cleavage of mismatch, and conformation sensitive gel electrophoresis. <i>Journal of Investigative Dermatology</i> , 1999 , 113, 673-86	4.3	63

15	A recurrent frameshift mutation in exon 19 of the type VII collagen gene (COL7A1) in Mexican patients with recessive dystrophic epidermolysis bullosa. <i>Experimental Dermatology</i> , 1999 , 8, 22-9	4	7
14	Molecular pathology of the cutaneous basement membrane zone. <i>Clinical and Experimental Dermatology</i> , 1999 , 24, 25-32	1.8	21
13	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex. <i>Human Mutation</i> , 1998 , 11, 279-85	4.7	36
12	Severe palmo-plantar hyperkeratosis in Dowling-Meara epidermolysis bullosa simplex caused by a mutation in the keratin 14 gene (KRT14). <i>Journal of Investigative Dermatology</i> , 1998 , 111, 893-5	4.3	36
11	Prognostic implications of determining 180 kDa bullous pemphigoid antigen (BPAG2) gene/protein pathology in neonatal junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , 1998 , 138, 661-6	4	10
10	Frameshift mutations in the type VII collagen gene (COL7A1) in five Mexican cousins with recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , 1998 , 138, 852-8	4	12
9	E210K mutation in the gene encoding the beta3 chain of laminin-5 (LAMB3) is predictive of a phenotype of generalized atrophic benign epidermolysis bullosa. <i>British Journal of Dermatology</i> , 1998 , 139, 325-31	4	20
8	A recurrent glycine substitution mutation, G2043R, in the type VII collagen gene (COL7A1) in dominant dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , 1998 , 139, 730-7	4	28
7	Pyloric atresia-junctional epidermolysis bullosa syndrome: mutations in the integrin beta4 gene (ITGB4) in two unrelated patients with mild disease. <i>British Journal of Dermatology</i> , 1998 , 139, 862-71	4	42
6	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex 1998 , 11, 279		3
5	Recurrent mutations in the type VII collagen gene (COL7A1) in patients with recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , 1997 , 109, 246-9	4.3	47
4	Recessive epidermolysis bullosa simplex associated with plectin mutations: infantile respiratory complications in two unrelated cases. <i>British Journal of Dermatology</i> , 1997 , 137, 898-906	4	7
3	A recurrent laminin 5 mutation in British patients with lethal (Herlitz) junctional epidermolysis bullosa: evidence for a mutational hotspot rather than propagation of an ancestral allele. <i>British Journal of Dermatology</i> , 1997 , 136, 674-677	4	1
2	Recessive epidermolysis bullosa simplex associated with plectin mutations: infantile respiratory complications in two unrelated cases. <i>British Journal of Dermatology</i> , 1997 , 137, 898-906	4	39
1	Recessive epidermolysis bullosa simplex associated with plectin mutations: infantile respiratory complications in two unrelated cases. <i>British Journal of Dermatology</i> , 1997 , 137, 898-906	4	9