

# J E Mellerio

## List of Publications by Year in descending order

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153  
papers

7,506  
citations

61984

43  
h-index

60623

81  
g-index

162  
all docs

162  
docs citations

162  
times ranked

4348  
citing authors

#	ARTICLE	IF	CITATIONS
1	Recommendations on pregnancy, childbirth and aftercare in epidermolysis bullosa: a consensus-based guideline*. British Journal of Dermatology, 2022, 186, 620-632.	1.5	4
2	Transcriptomic profiling of recessive dystrophic epidermolysis bullosa wounded skin highlights drug repurposing opportunities to improve wound healing. Experimental Dermatology, 2022, 31, 420-426.	2.9	9
3	Epidermolysis bullosa acquisita: a case series of three paediatric patients. Clinical and Experimental Dermatology, 2022, , .	1.3	1
4	The epidemiology of epidermolysis bullosa in England and Wales: data from the national epidermolysis bullosa database*. British Journal of Dermatology, 2022, 186, 843-848.	1.5	22
5	Heterogeneous addiction to transforming growth factor-β signalling in recessive dystrophic epidermolysis bullosa-associated cutaneous squamous cell carcinoma*. British Journal of Dermatology, 2021, 184, 697-708.	1.5	12
6	Prevalence, pathophysiology and management of itch in epidermolysis bullosa*. British Journal of Dermatology, 2021, 184, 816-825.	1.5	38
7	Characteristics of children with Netherton syndrome: a review of 21 patients. Journal of the European Academy of Dermatology and Venereology, 2021, 35, e466-e469.	2.4	4
8	Practical management of epidermolysis bullosa: consensus clinical position statement from the European Reference Network for Rare Skin Diseases. Journal of the European Academy of Dermatology and Venereology, 2021, 35, 2349-2360.	2.4	12
9	Otological complications in inversa type recessive dystrophic epidermolysis bullosa. Clinical and Experimental Dermatology, 2021, , .	1.3	0
10	Genotype-phenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. British Journal of Dermatology, 2020, 182, 729-737.	1.5	47
11	Foot care in epidermolysis bullosa: evidence-based guideline. British Journal of Dermatology, 2020, 182, 593-604.	1.5	17
12	EBGene trial: patient preselection outcomes for the European GENEGRIFT <i>ex vivo</i> phase I/II gene therapy trial for recessive dystrophic epidermolysis bullosa. British Journal of Dermatology, 2020, 182, 794-797.	1.5	19
13	The psychological functioning of children with epidermolysis bullosa and its relationship with specific aspects of disease. British Journal of Dermatology, 2020, 182, 789-790.	1.5	3
14	Pseudoporphyria induced by ultraviolet radiation. Australasian Journal of Dermatology, 2020, 61, 177-179.	0.7	0
15	Homozygous Nonsense Mutation in DSC3 Resulting in Skin Fragility and Hypotrichosis. Journal of Investigative Dermatology, 2020, 140, 1285-1288.	0.7	8
16	Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. British Journal of Dermatology, 2020, 183, 614-627.	1.5	406
17	PLACK syndrome: the penny dropped. Clinical and Experimental Dermatology, 2020, 45, 1091-1092.	1.3	2
18	Meeting Report: The First Global Congress on Epidermolysis Bullosa, EB2020 London: Toward Treatment and Cure. Journal of Investigative Dermatology, 2020, 140, 1681-1687.	0.7	11

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19	Enduring support for the case report. <i>British Journal of Dermatology</i> , 2019, 181, 429-430.	1.5	2
20	Beta blockers for infantile haemangiomas: where should we go from here?. <i>British Journal of Dermatology</i> , 2019, 180, 450-451.	1.5	2
21	Potential therapeutic targeting of inflammation in epidermolysis bullosa simplex. <i>British Journal of Dermatology</i> , 2019, 180, 258-260.	1.5	3
22	Itch and scratch: could pain be the catch?. <i>British Journal of Dermatology</i> , 2018, 179, 1029-1029.	1.5	0
23	Predictive phenotyping of inherited ichthyosis by next-generation DNA sequencing. <i>British Journal of Dermatology</i> , 2017, 176, 249-251.	1.5	8
24	Focal dermal hypoplasia: inheritance from father to daughter. <i>Clinical and Experimental Dermatology</i> , 2017, 42, 457-459.	1.3	4
25	Wound healing in epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2017, 177, e193-e195.	1.5	8
26	Neonatal aggressive systemic mastocytosis. <i>British Journal of Dermatology</i> , 2017, 177, 1167-1168.	1.5	0
27	Considerations in surgical management of a Buschke-Lowenstein tumor in Netherton syndrome: A case report. <i>Pediatric Dermatology</i> , 2017, 34, e328-e330.	0.9	11
28	G413(P)...Should the frequency of echocardiogram screening be increased in severe subtypes of epidermolysis bullosa?. , 2017, , .		0
29	Autosomal dominant diffuse nonepidermolytic palmoplantar keratoderma due to a recurrent mutation in aquaporin-5. <i>British Journal of Dermatology</i> , 2016, 174, 430-432.	1.5	8
30	Mutations in <i>EXPH5</i> underlie a rare subtype of autosomal recessive epidermolysis bullosa simplex. <i>British Journal of Dermatology</i> , 2016, 174, 452-453.	1.5	9
31	Early-onset dermatosis papulosa nigra. <i>British Journal of Dermatology</i> , 2016, 174, 1148-1150.	1.5	6
32	Ichthyosis Prematurity Syndrome. <i>JAMA Dermatology</i> , 2016, 152, 1055.	4.1	8
33	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.	1.5	102
34	Mesenchymal stem cell therapy for recessive dystrophic epidermolysis bullosa: prospects and clinical progress. <i>Expert Opinion on Orphan Drugs</i> , 2016, 4, 343-345.	0.8	2
35	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.	0.7	119
36	Pityriasis rubra pilaris with histologic features of lichen nitidus. <i>Journal of the American Academy of Dermatology</i> , 2015, 73, 336-337.	1.2	5

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37	Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. <i>British Journal of Dermatology</i> , 2015, 172, 94-100.	1.5	74
38	Clinical features and <i>WNT10A</i> mutations in seven unrelated cases of Schöpf-Schulz-Passarge syndrome. <i>British Journal of Dermatology</i> , 2014, 171, 1211-1214.	1.5	24
39	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.	0.7	24
40	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.	1.9	1
41	Mutations in <i>EXPH5</i> result in autosomal recessive inherited skin fragility. <i>British Journal of Dermatology</i> , 2014, 170, 196-199.	1.5	13
42	Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification. <i>Journal of the American Academy of Dermatology</i> , 2014, 70, 1103-1126.	1.2	747
43	Under-recognition of acral peeling skin syndrome: 59 new cases with 15 novel mutations. <i>British Journal of Dermatology</i> , 2014, 171, 1206-1210.	1.5	28
44	Epithelial Inflammation Resulting from an Inherited Loss-of-Function Mutation in EGFR. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2570-2578.	0.7	71
45	Pain, purpura and curly hairs. <i>Clinical and Experimental Dermatology</i> , 2013, 38, 940-942.	1.3	8
46	Serum levels of high mobility group box 1 correlate with disease severity in recessive dystrophic epidermolysis bullosa. <i>Experimental Dermatology</i> , 2013, 22, 433-435.	2.9	30
47	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, 1025-1033.	1.5	111
48	Phase I Study Protocol for <i>Ex Vivo</i> Lentiviral Gene Therapy for the Inherited Skin Disease, Netherton Syndrome. <i>Human Gene Therapy Clinical Development</i> , 2013, 24, 182-190.	3.1	37
49	Homozygous variegate porphyria presenting with developmental and language delay in childhood. <i>Clinical and Experimental Dermatology</i> , 2013, 38, 737-740.	1.3	13
50	Epidermolysis Bullosa and Chronic Wounds. <i>Advances in Skin and Wound Care</i> , 2013, 26, 189-190.	1.0	0
51	Immunofluorescence antigen mapping for hereditary epidermolysis bullosa. <i>Indian Journal of Dermatology, Venereology and Leprology</i> , 2012, 78, 692.	0.6	19
52	Autosomal Recessive Epidermolysis Bullosa Simplex Due to Loss of BPAG1-e Expression. <i>Journal of Investigative Dermatology</i> , 2012, 132, 742-744.	0.7	55
53	Fluoroscopically Guided Dilatation of Esophageal Strictures in Patients With Dystrophic Epidermolysis Bullosa: Long-Term Results. <i>American Journal of Roentgenology</i> , 2012, 199, 208-212.	2.2	28
54	A consensus approach to wound care in epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , 2012, 67, 904-917.	1.2	148

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55	Gastrostomy Tube Feeding in Children with Epidermolysis Bullosa: Consideration of Key Issues. <i>Pediatric Dermatology</i> , 2012, 29, 277-284.	0.9	22
56	A pyrexial unwell child with a papular eruption. <i>Clinical and Experimental Dermatology</i> , 2012, 37, 811-813.	1.3	3
57	Corrigendum to "Novel and recurrent COL7A1 mutations in Chilean patients with dystrophic epidermolysis bullosa" [J. Dermatol. Sci. 65 (2012) 149-152]. <i>Journal of Dermatological Science</i> , 2012, 66, 85.	1.9	0
58	Novel and recurrent COL7A1 mutations in Chilean patients with dystrophic epidermolysis bullosa. <i>Journal of Dermatological Science</i> , 2012, 65, 149-152.	1.9	16
59	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. <i>American Journal of Human Genetics</i> , 2012, 91, 1115-1121.	6.2	65
60	Desmosomal genodermatoses. <i>British Journal of Dermatology</i> , 2012, 166, 36-45.	1.5	74
61	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, 134-144.	1.5	27
62	MBTPS2 mutation in a British pedigree with keratosis follicularis spinulosa decalvans. <i>Clinical and Experimental Dermatology</i> , 2012, 37, 631-634.	1.3	16
63	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.	2.3	3
64	Recalcitrant generalized eruption and low alkaline phosphatase: think zinc. <i>Clinical and Experimental Dermatology</i> , 2011, 36, 225-226.	1.3	1
65	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, 579-584.	1.3	34
66	An unusual case of epidermolysis bullosa complicated by persistent oligoarticular juvenile idiopathic arthritis; lessons to be learned. <i>Pediatric Rheumatology</i> , 2011, 9, 13.	2.1	4
67	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, 1100-1107.	2.5	74
68	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, 160-167.	3.2	35
69	Harlequin Ichthyosis. <i>Archives of Dermatology</i> , 2011, 147, 681.	1.4	145
70	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, 1771-1774.	0.7	59
71	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, 262-266.	1.3	41
72	Dilated Cardiomyopathy in Epidermolysis Bullosa: A Retrospective, Multicenter Study. <i>Pediatric Dermatology</i> , 2010, 27, 238-243.	0.9	37

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73	Molecular basis of EEC (ectrodactyly, ectodermal dysplasia, clefting) syndrome: five new mutations in the DNA-binding domain of the <i>TP63</i> gene and genotype-phenotype correlation. <i>British Journal of Dermatology</i> , 2010, 162, 201-207.	1.5	36
74	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, 624-629.	1.5	36
75	Bullous pemphigoid in a patient with suspected non-Herlitz junctional epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 2010, 35, 881-884.	1.3	4
76	Revertant Mosaicism in Recessive Dystrophic Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , 2010, 130, 1937-1940.	0.7	55
77	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, 1551-1557.	0.7	136
78	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, 1543-1550.	0.7	49
79	Ectodermal Dysplasia-Skin Fragility Syndrome. <i>Dermatologic Clinics</i> , 2010, 28, 125-129.	1.7	64
80	Epidermolysis Bullosa Care in the United Kingdom. <i>Dermatologic Clinics</i> , 2010, 28, 395-396.	1.7	9
81	Infection and Colonization in Epidermolysis Bullosa. <i>Dermatologic Clinics</i> , 2010, 28, 267-269.	1.7	51
82	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, 709-711.	0.6	46
83	Osteopenia and Osteoporosis in Epidermolysis Bullosa. <i>Dermatologic Clinics</i> , 2010, 28, 353-355.	1.7	24
84	Genitourinary Tract Involvement in Epidermolysis Bullosa. <i>Dermatologic Clinics</i> , 2010, 28, 343-346.	1.7	21
85	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.	1.3	40
86	Autosomal dominant junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2009, 160, 1094-1097.	1.5	33
87	PORCN gene mutations and the protean nature of focal dermal hypoplasia. <i>British Journal of Dermatology</i> , 2009, 160, 1103-1109.	1.5	33
88	Epidermolysis bullosa pruriginosa in association with lichen planopilaris. <i>Clinical and Experimental Dermatology</i> , 2009, 34, e825-e828.	1.3	10
89	Maxillary alveolar process fracture complicating intubation in a patient with epidermolysis bullosa. <i>Paediatric Anaesthesia</i> , 2009, 19, 706-707.	1.1	5
90	Extracutaneous manifestations and complications of inherited epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , 2009, 61, 367-384.	1.2	234

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91	Extracutaneous manifestations and complications of inherited epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , 2009, 61, 387-402.	1.2	218
92	Novel and recurring ABCA12 mutations associated with harlequin ichthyosis: implications for prenatal diagnosis. <i>British Journal of Dermatology</i> , 2008, 158, 611-613.	1.5	28
93	Potential of Fibroblast Cell Therapy for Recessive Dystrophic Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , 2008, 128, 2179-2189.	0.7	219
94	Congenital Anetoderma in a Preterm Infant. <i>Pediatric Dermatology</i> , 2008, 25, 626-629.	0.9	12
95	Gastrointestinal complications of epidermolysis bullosa in children. <i>British Journal of Dermatology</i> , 2008, 158, 1308-1314.	1.5	110
96	Anaesthetic management of two different modes of delivery in patients with dystrophic epidermolysis bullosa. <i>International Journal of Obstetric Anesthesia</i> , 2008, 17, 153-158.	0.4	27
97	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, 931-950.	1.2	812
98	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, 385-391.	1.0	29
99	A spectrum of mutations in keratins K6a, K16 and K17 causing pachyonychia congenita. <i>Journal of Dermatological Science</i> , 2007, 48, 199-205.	1.9	49
100	Plectin defects in epidermolysis bullosa simplex with muscular dystrophy. <i>Muscle and Nerve</i> , 2007, 35, 24-35.	2.2	60
101	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.	0.7	47
102	Oral 5, Expanding choice for prenatal testing in couples at reproductive risk of Herlitz junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2007, 156, 1404-1404.	1.5	0
103	Generalized pustular eruption in a 5-year-old boy. <i>Clinical and Experimental Dermatology</i> , 2007, 33, 79-80.	1.3	2
104	Prenatal diagnosis for severe inherited skin disorders: 25 years' experience. <i>British Journal of Dermatology</i> , 2006, 154, 106-113.	1.5	86
105	Bone mineralization in children with epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2006, 154, 959-962.	1.5	56
106	Target proteins in inherited and acquired blistering skin disorders. <i>Clinical and Experimental Dermatology</i> , 2006, 31, 252-259.	1.3	35
107	An unusual patient with Rothmund-Thomson syndrome, porokeratosis and bilateral iris dysgenesis. <i>Clinical and Experimental Dermatology</i> , 2006, 31, 401-403.	1.3	15
108	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, 2039-2043.	0.7	33



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109	Epidermolysis bullosa. <i>British Journal of Hospital Medicine</i> (London, England: 2005), 2006, 67, 188-191.	0.5	21
110	Histopathological features of gastrointestinal mucosal biopsy specimens in children with epidermolysis bullosa. <i>Journal of Clinical Pathology</i> , 2006, 60, 843-844.	2.0	8
111	Thalidomide in the management of epidermolysis bullosa pruriginosa. <i>British Journal of Dermatology</i> , 2005, 152, 1332-1334.	1.5	58
112	Complete paternal uniparental isodisomy of chromosome 1 resulting in Herlitz junctional epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 2005, 30, 71-74.	1.3	36
113	Multiple dermatofibromas associated with lupus profundus. <i>Clinical and Experimental Dermatology</i> , 2005, 30, 128-130.	1.3	12
114	Prenatal diagnosis of Herlitz junctional epidermolysis bullosa in nonidentical twins. <i>Clinical and Experimental Dermatology</i> , 2005, 30, 180-182.	1.3	7
115	Neonatal diagnosis of Kindler syndrome. <i>Journal of Dermatological Science</i> , 2005, 39, 183-185.	1.9	22
116	ADULT ectodermal dysplasia syndrome resulting from the missense mutation R298Q in the p63 gene. <i>Clinical and Experimental Dermatology</i> , 2004, 29, 669-672.	1.3	29
117	Dilemmas in distinguishing between dominant and recessive forms of dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , 2003, 149, 810-818.	1.5	31
118	An unusual N-terminal deletion of the laminin $\hat{A}3a$ isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. <i>Human Molecular Genetics</i> , 2003, 13, 365-365.	2.9	81
119	An unusual N-terminal deletion of the laminin $\hat{A}3a$ isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. <i>Human Molecular Genetics</i> , 2003, 12, 2395-2409.	2.9	123
120	Skin disease in Gulf war veterans. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2002, 95, 671-676.	0.5	6
121	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, 833-840.	2.9	246
122	alpha6beta4 integrin abnormalities in junctional epidermolysis bullosa with pyloric atresia. <i>British Journal of Dermatology</i> , 2001, 144, 408-414.	1.5	65
123	Three cases of de novo dominant dystrophic epidermolysis bullosa associated with the mutation G2043R in COL7A1. <i>Clinical and Experimental Dermatology</i> , 2001, 26, 97-99.	1.3	19
124	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , 2001, 26, 93-96.	1.3	11
125	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , 2001, 26, 93-96.	1.3	13
126	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , 2001, 26, 93-6.	1.3	22



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127	Heterogeneous Mutations of the Type VII Collagen Gene (COL7A1) in Dystrophic Epidermolysis Bullosa. <i>Clinical Science</i> , 2000, 98, 18P-18P.	0.0	0
128	Heterozygous germline missense mutation in the p63 gene underlying EEC syndrome. <i>Clinical and Experimental Dermatology</i> , 2000, 25, 441-443.	1.3	38
129	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, 314-321.	0.7	67
130	Skin fragility and hypohidrotic ectodermal dysplasia resulting from ablation of plakophilin 1. <i>British Journal of Dermatology</i> , 1999, 140, 297-307.	1.5	98
131	A recurrent COL7A1 mutation, R2814X, in British patients with recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 1999, 24, 37-39.	1.3	12
132	Frequency of the CCR5 gene 32-basepair deletion in Hispanic Mexicans. <i>Clinical and Experimental Dermatology</i> , 1999, 24, 127-129.	1.3	5
133	Recurrent molecular abnormalities in type VII collagen in southern Italian patients with recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , 1999, 24, 232-235.	1.3	12
134	Allelic Heterogeneity of Dominant and Recessive COL7A1 Mutations Underlying Epidermolysis Bullosa Pruriginosa. <i>Journal of Investigative Dermatology</i> , 1999, 112, 984-987.	0.7	82
135	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, 673-686.	0.7	76
136	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, 22-29.	2.9	9
137	Molecular pathology of the cutaneous basement membrane zone. <i>Clinical and Experimental Dermatology</i> , 1999, 24, 25-32.	1.3	24
138	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex. <i>Human Mutation</i> , 1998, 11, 279-285.	2.5	41
139	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, 893-895.	0.7	43
140	Prognostic implications of determining 180kDa bullous pemphigoid antigen (BPAG2) gene/protein pathology in neonatal junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , 1998, 138, 661-666.	1.5	12
141	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, 852-858.	1.5	17
142	E210K mutation in the gene encoding the $\beta$ 3 chain of laminin-5 (LAMB3) is predictive of a phenotype of generalized atrophic benign epidermolysis bullosa. <i>British Journal of Dermatology</i> , 1998, 139, 325-331.	1.5	27
143	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, 730-737.	1.5	36
144	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, 862-871.	1.5	50

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145	Plakophilin 1: Partial genomic organisation and mutations resulting in ectodermal dysplasia/skin fragility syndrome. <i>Journal of Dermatological Science</i> , 1998, 16, S1.	1.9	0
146	Allelic heterogeneity of type VII collagen gene (COL7A1) mutations in epidermolysis bullosa pruriginosa. <i>Journal of Dermatological Science</i> , 1998, 16, S36.	1.9	0
147	Type VII collagen gene (COL7A1) mutation screening in recessive dystrophic epidermolysis bullosa using the protein truncation test. <i>Journal of Dermatological Science</i> , 1998, 16, S146.	1.9	0
148	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex. <i>Human Mutation</i> , 1998, 11, 279-285.	2.5	4
149	Recurrent Mutations in the Type VII Collagen Gene (COL7A1) in Patients with Recessive Dystrophic Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , 1997, 109, 246-249.	0.7	52
150	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.	1.5	25
151	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.	1.5	3
152	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.	1.5	43
153	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.	1.5	10