Giovanna Zambruno

List of Publications by Citations

224 papers	10,393 citations	54 h-index	96 g-index
233 ext. papers	12,034 ext. citations	4.1 avg, IF	5.52 L-index

#	Paper	IF	Citations
224	The classification of inherited epidermolysis bullosa (EB): Report of the Third International Consensus Meeting on Diagnosis and Classification of EB. *Journal of the American Academy of Dermatology*, **2008**, 58, 931-50	4.5	690
223	Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. *Journal of the American Academy of Dermatology*, **2014**, 70, 1103-26	4.5	596
222	Spink5-deficient mice mimic Netherton syndrome through degradation of desmoglein 1 by epidermal protease hyperactivity. *Nature Genetics*, **2005**, 37, 56-65	36.3	300
221	Transforming growth factor-beta 1 modulates beta 1 and beta 5 integrin receptors and induces the de novo expression of the alpha v beta 6 heterodimer in normal human keratinocytes: implications for wound healing. *Journal of Cell Biology*, **1995**, 129, 853-65	7.3	296
220	The control of epidermal stem cells (holoclones) in the treatment of massive full-thickness burns with autologous keratinocytes cultured on fibrin. *Transplantation*, **1999**, 68, 868-79	1.8	283
219	Mutations in the C7orf11 (TTDN1) gene in six nonphotosensitive trichothiodystrophy patients: no obvious genotype-phenotype relationships. *Human Mutation*, **2007**, 28, 92-6	4.7	237
218	TFIIH-dependent MMP-1 overexpression in trichothiodystrophy leads to extracellular matrix alterations in patient skin. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 1499-504	11.5	221
217	Involvement of the mismatch repair system in temozolomide-induced apoptosis. *Molecular Pharmacology*, **1998**, 54, 334-41	4.3	206
216	New functions of XPC in the protection of human skin cells from oxidative damage. *EMBO Journal*, **2006**, 25, 4305-15	13	204
215	Definitions and outcome measures for bullous pemphigoid: recommendations by an international panel of experts. *Journal of the American Academy of Dermatology*, **2012**, 66, 479-85	4.5	203
214	Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *British Journal of Dermatology*, **2020**, 183, 614-627	4	179
213	Management of bullous pemphigoid: the European Dermatology Forum consensus in collaboration with the European Academy of Dermatology and Venereology. *British Journal of Dermatology*, **2015** , 172, 867-77	4	178
212	Mutations in PYCR1 cause cutis laxa with progeroid features. *Nature Genetics*, **2009**, 41, 1016-21	36.3	178
211	Pemphigus. S2 Guideline for diagnosis and treatment--guided by the European Dermatology Forum (EDF) in cooperation with the European Academy of Dermatology and Venereology (EADV). *Journal of the European Academy of Dermatology and Venereology*, **2015**, 29, 405-14	4.6	165
210	Distinctive integrin expression in the newly forming epidermis during wound healing in humans. *Journal of Investigative Dermatology*, **1993**, 101, 600-4	4.3	161
209	Corneodesmosomal cadherins are preferential targets of stratum corneum trypsin- and chymotrypsin-like hyperactivity in Netherton syndrome. *Journal of Investigative Dermatology*, **2006**, 126, 1622-32	4.3	148
208	Mice overexpressing placenta growth factor exhibit increased vascularization and vessel permeability. *Journal of Cell Science*, **2002**, 115, 2559-2567	5.3	144

207	Multicenter prospective study of the humoral autoimmune response in bullous pemphigoid. *Clinical Immunology*, **2008**, 128, 415-26	9	141
206	A homozygous mutation in the integrin alpha6 gene in junctional epidermolysis bullosa with pyloric atresia. *Journal of Clinical Investigation*, **1997**, 99, 2826-31	15.9	140
205	Distinct vascular endothelial growth factor signals for lymphatic vessel enlargement and sprouting. *Journal of Experimental Medicine*, **2007**, 204, 1431-40	16.6	137
204	Netherton syndrome: disease expression and spectrum of SPINK5 mutations in 21 families. *Journal of Investigative Dermatology*, **2002**, 118, 352-61	4.3	136
203	Mice overexpressing placenta growth factor exhibit increased vascularization and vessel permeability. *Journal of Cell Science*, **2002**, 115, 2559-67	5.3	135
202	Diabetes impairs adipose tissue-derived stem cell function and efficiency in promoting wound healing. *Wound Repair and Regeneration*, **2013**, 21, 545-53	3.6	133
201	Human melanoma cells secrete and respond to placenta growth factor and vascular endothelial growth factor. *Journal of Investigative Dermatology*, **2000**, 115, 1000-7	4.3	128
200	The role of CSA in the response to oxidative DNA damage in human cells. *Oncogene*, **2007**, 26, 4336-43	9.2	126
199	Pemphigus autoantibodies generated through somatic mutations target the desmoglein-3 cis-interface. *Journal of Clinical Investigation*, **2012**, 122, 3781-90	15.9	112
198	Corrective transduction of human epidermal stem cells in laminin-5-dependent junctional epidermolysis bullosa. *Human Gene Therapy*, **1998**, 9, 1359-70	4.8	105
197	Adenovirus-mediated VEGF(165) gene transfer enhances wound healing by promoting angiogenesis in CD1 diabetic mice. *Gene Therapy*, **2002**, 9, 1271-7	4	101
196	Demonstration of epitope-spreading phenomena in bullous pemphigoid: results of a prospective multicenter study. *Journal of Investigative Dermatology*, **2011**, 131, 2271-80	4.3	97
195	Bullous pemphigoid: from the clinic to the bench. *Clinics in Dermatology*, **2012**, 30, 3-16	3	96
194	Placenta growth factor in diabetic wound healing: altered expression and therapeutic potential. *American Journal of Pathology*, **2006**, 169, 1167-82	5.8	96
193	Definitions and outcome measures for mucous membrane pemphigoid: recommendations of an international panel of experts. *Journal of the American Academy of Dermatology*, **2015**, 72, 168-74	4.5	93
192	Vascular endothelial growth factor receptor-1 is deposited in the extracellular matrix by endothelial cells and is a ligand for the alpha 5 beta 1 integrin. *Journal of Cell Science*, **2003**, 116, 3479-89	5.3	86
191	Increased melanoma growth and metastasis spreading in mice overexpressing placenta growth factor. *American Journal of Pathology*, **2006**, 169, 643-54	5.8	83
190	Kindler syndrome: extension of FERMT1 mutational spectrum and natural history. *Human Mutation*, **2011**, 32, 1204-12	4.7	82

189	Mutations in PVRL4, encoding cell adhesion molecule nectin-4, cause ectodermal dysplasia-syndactyly syndrome. *American Journal of Human Genetics*, **2010**, 87, 265-73	11	78
188	Vascular endothelial growth factor-C expression correlates with lymph node localization of human melanoma metastases. *Cancer*, **2003**, 98, 789-97	6.4	78
187	Multicentre consensus recommendations for skin care in inherited epidermolysis bullosa. *Orphanet Journal of Rare Diseases*, **2014**, 9, 76	4.2	76
186	TSH receptor and thyroid-specific gene expression in human skin. *Journal of Investigative Dermatology*, **2010**, 130, 93-101	4.3	75
185	Placenta growth factor is induced in human keratinocytes during wound healing. *Journal of Investigative Dermatology*, **2000**, 115, 388-95	4.3	75
184	Characterization of the anti-BP180 autoantibody reactivity profile and epitope mapping in bullous pemphigoid patients. *Journal of Investigative Dermatology*, **2004**, 122, 103-10	4.3	71
183	The 420K LEKTI variant alters LEKTI proteolytic activation and results in protease deregulation: implications for atopic dermatitis. *Human Molecular Genetics*, **2012**, 21, 4187-200	5.6	67
182	Fine mapping of the PSORS4 psoriasis susceptibility region on chromosome 1q21. *Journal of Investigative Dermatology*, **2001**, 116, 728-30	4.3	65
181	The international dystrophic epidermolysis bullosa patient registry: an online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. *Human Mutation*, **2011**, 32, 1100-7	4.7	63
180	Granulocyte/macrophage colony-stimulating factor treatment of human chronic ulcers promotes angiogenesis associated with de novo vascular endothelial growth factor transcription in the ulcer bed. *British Journal of Dermatology*, **2006**, 154, 34-41	4	62
179	Evidence for differential S100 gene over-expression in psoriatic patients from genetically heterogeneous pedigrees. *Human Genetics*, **2002**, 111, 310-3	6.3	62
178	Monozygotic twins discordant for recessive dystrophic epidermolysis bullosa phenotype highlight the role of TGF-β signalling in modifying disease severity. *Human Molecular Genetics*, **2014**, 23, 3907-22	5.6	61
177	Quality of life in patients with epidermolysis bullosa. *British Journal of Dermatology*, **2009**, 161, 869-77	4	61
176	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
175	Updated S2K guidelines on the management of pemphigus vulgaris and foliaceus initiated by the european academy of dermatology and venereology (EADV). *Journal of the European Academy of Dermatology and Venereology*, **2020**, 34, 1900-1913	4.6	60
174	Development of a novel ELISA system for detection of anti-BP180 IgG and characterization of autoantibody profile in bullous pemphigoid patients. *British Journal of Dermatology*, **2004**, 151, 1004-10	4	59
173	Stromal microenvironment in type VII collagen-deficient skin: The ground for squamous cell carcinoma development. *Matrix Biology*, **2017**, 63, 1-10	11.4	58
172	Long-term engraftment of single genetically modified human epidermal holoclones enables safety pre-assessment of cutaneous gene therapy. *Molecular Therapy*, **2007**, 15, 1670-6	11.7	57

171	The placenta growth factor in skin angiogenesis. *Journal of Dermatological Science*, **2006**, 41, 11-9	4.3	54
170	Calculation of cut-off values based on the Autoimmune Bullous Skin Disorder Intensity Score (ABSIS) and Pemphigus Disease Area Index (PDAI) pemphigus scoring systems for defining moderate, significant and extensive types of pemphigus. *British Journal of Dermatology*, **2016**, 175, 142-9	4	52
169	Monoallelic Mutations in the Translation Initiation Codon of KLHL24 Cause Skin Fragility. *American Journal of Human Genetics*, **2016**, 99, 1395-1404	11	51
168	Interleukin-22 Promotes Wound Repair in Diabetes by Improving Keratinocyte Pro-Healing Functions. *Journal of Investigative Dermatology*, **2015**, 135, 2862-2870	4.3	50
167	Bmi-1 reduction plays a key role in physiological and premature aging of primary human keratinocytes. *Journal of Investigative Dermatology*, **2010**, 130, 1048-62	4.3	50
166	Genotype-phenotype correlation in italian patients with dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **2002**, 119, 1456-62	4.3	49
165	VLA protein expression on epidermal cells (keratinocytes, Langerhans cells, melanocytes): a light and electron microscopic immunohistochemical study. *British Journal of Dermatology*, **1991**, 124, 135-45	4	49
164	Sirtinol treatment reduces inflammation in human dermal microvascular endothelial cells. *PLoS ONE*, **2011**, 6, e24307	3.7	49
163	Differential role of transcription-coupled repair in UVB-induced response of human fibroblasts and keratinocytes. *Cancer Research*, **2005**, 65, 432-8	10.1	49
162	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. *American Journal of Human Genetics*, **2015**, 97, 99-110	11	48
161	Toward epidermal stem cell-mediated ex vivo gene therapy of junctional epidermolysis bullosa. *Human Gene Therapy*, **2000**, 11, 2283-7	4.8	48
160	Proteolytic activation cascade of the Netherton syndrome-defective protein, LEKTI, in the epidermis: implications for skin homeostasis. *Journal of Investigative Dermatology*, **2011**, 131, 2223-32	4.3	47
159	Molecular basis of Kindler syndrome in Italy: novel and recurrent Alu/Alu recombination, splice site, nonsense, and frameshift mutations in the KIND1 gene. *Journal of Investigative Dermatology*, **2006**, 126, 1776-83	4.3	47
158	Apoptosis and efficient repair of DNA damage protect human keratinocytes against UVB. *Cell Death and Differentiation*, **2003**, 10, 754-6	12.7	45
157	In vitro antitumour activity of resveratrol in human melanoma cells sensitive or resistant to temozolomide. *Melanoma Research*, **2004**, 14, 189-96	3.3	45
156	Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. *British Journal of Dermatology*, **2020**, 182, 574-592	4	45
155	Binding properties, cell delivery, and gene transfer of adenoviral penton base displaying bacteriophage. *Virology*, **2001**, 282, 102-12	3.6	44
154	Oral pemphigoid autoantibodies preferentially target BP180 ectodomain. *Clinical Immunology*, **2007**, 122, 207-13	9	42

153	The first COL7A1 mutation survey in a large Spanish dystrophic epidermolysis bullosa cohort: c.6527insC disclosed as an unusually recurrent mutation. *British Journal of Dermatology*, **2010**, 163, 155-61	4	41
152	The intracellular and extracellular domains of BP180 antigen comprise novel epitopes targeted by pemphigoid gestationis autoantibodies. *Journal of Investigative Dermatology*, **2007**, 127, 864-73	4.3	40
151	Inactivation of p16INK4a (inhibitor of cyclin-dependent kinase 4A) immortalizes primary human keratinocytes by maintaining cells in the stem cell compartment. *FASEB Journal*, **2006**, 20, 1516-8	0.9	39
150	Efficiency of translation termination in humans is highly dependent upon nucleotides in the neighbourhood of a (premature) termination codon. *Journal of Medical Genetics*, **2011**, 48, 640-4	5.8	36
149	Pathomechanisms of Altered Wound Healing in Recessive Dystrophic Epidermolysis Bullosa. *American Journal of Pathology*, **2017**, 187, 1445-1453	5.8	34
148	Rituximab immunotherapy in pemphigus: therapeutic effects beyond B-cell depletion. *Journal of Investigative Dermatology*, **2008**, 128, 2745-7	4.3	34
147	Large International Validation of ABSIS and PDAI Pemphigus Severity Scores. *Journal of Investigative Dermatology*, **2019**, 139, 31-37	4.3	33
146	Epidermolysis Bullosa-Associated Squamous Cell Carcinoma: From Pathogenesis to Therapeutic Perspectives. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	32
145	Compound heterozygosity for a recessive glycine substitution and a splice site mutation in the COL7A1 gene causes an unusually mild form of localized recessive dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **1998**, 111, 744-50	4.3	30
144	A homozygous nonsense mutation in type XVII collagen gene (COL17A1) uncovers an alternatively spliced mRNA accounting for an unusually mild form of non-Herlitz junctional epidermolysis bullosa. *Journal of Investigative Dermatology*, **2001**, 116, 182-7	4.3	29
143	Characterization of the loricrin (LOR) gene as a positional candidate for the PSORS4 psoriasis susceptibility locus. *Annals of Human Genetics*, **2004**, 68, 639-45	2.2	28
142	Prevalence of collagen VII-specific autoantibodies in patients with autoimmune and inflammatory diseases. *BMC Immunology*, **2012**, 13, 16	3.7	27
141	Concomitant activation of Wnt pathway and loss of mismatch repair function in human melanoma. *Genes Chromosomes and Cancer*, **2008**, 47, 614-24	5	26
140	High-frequency microsatellite instability is associated with defective DNA mismatch repair in human melanoma. *Journal of Investigative Dermatology*, **2002**, 118, 79-86	4.3	26
139	A glutamine insertion in the 1A alpha helical domain of the keratin 4 gene in a familial case of white sponge nevus. *Journal of Investigative Dermatology*, **2000**, 114, 388-91	4.3	26
138	Family burden in epidermolysis bullosa is high independent of disease type/subtype. *Acta Dermato-Venereologica*, **2010**, 90, 607-11	2.2	25
137	A proangiogenic peptide derived from vascular endothelial growth factor receptor-1 acts through alpha5beta1 integrin. *Blood*, **2008**, 111, 3479-88	2.2	25
136	Dystrophic epidermolysis bullosa pruriginosa in Italy: clinical and molecular characterization. *Clinical Genetics*, **2006**, 70, 339-47	4	25

135	SPINK5, the defective gene in netherton syndrome, encodes multiple LEKTI isoforms derived from alternative pre-mRNA processing. *Journal of Investigative Dermatology*, **2006**, 126, 315-24	4.3	25
134	Laminin-5 mutational analysis in an Italian cohort of patients with junctional epidermolysis bullosa. *Journal of Investigative Dermatology*, **2004**, 123, 639-48	4.3	25
133	Compound heterozygosity for an out-of-frame deletion and a splice site mutation in the LAMB3 gene causes nonlethal junctional epidermolysis bullosa. *Biochemical and Biophysical Research Communications*, **1998**, 243, 758-64	3.4	25
132	Decorin counteracts disease progression in mice with recessive dystrophic epidermolysis bullosa. *Matrix Biology*, **2019**, 81, 3-16	11.4	25
131	Nectin-4 mutations causing ectodermal dysplasia with syndactyly perturb the rac1 pathway and the kinetics of adherens junction formation. *Journal of Investigative Dermatology*, **2014**, 134, 2146-2153	4.3	24
130	Clinical and genetic heterogeneity in keratosis follicularis spinulosa decalvans. *European Journal of Medical Genetics*, **2009**, 52, 53-8	2.6	23
129	Novel mutations in the LAMC2 gene in non-Herlitz junctional epidermolysis bullosa: effects on laminin-5 assembly, secretion, and deposition. *Journal of Investigative Dermatology*, **2001**, 117, 731-9	4.3	23
128	Expression of an estrogen receptor-associated protein (p29) in epithelial tumors of the skin. *Journal of Cutaneous Pathology*, **1989**, 16, 272-6	1.7	23
127	Hereditary palmoplantar keratodermas. Part I. Non-syndromic palmoplantar keratodermas: classification, clinical and genetic features. *Journal of the European Academy of Dermatology and Venereology*, **2018**, 32, 704-719	4.6	22
126	Kindlin-1 regulates integrin dynamics and adhesion turnover. *PLoS ONE*, **2013**, 8, e65341	3.7	22
125	Ichthyosis with confetti: clinics, molecular genetics and management. *Orphanet Journal of Rare Diseases*, **2015**, 10, 115	4.2	21
124	Expression of the soluble vascular endothelial growth factor receptor-1 in cutaneous melanoma: role in tumour progression. *British Journal of Dermatology*, **2011**, 164, 1061-70	4	21
123	Genotype-phenotype relationships in trichothiodystrophy patients with novel splicing mutations in the XPD gene. *Human Mutation*, **2009**, 30, 438-45	4.7	21
122	Immunofluorescence analysis of villous trophoblasts: a tool for prenatal diagnosis of inherited epidermolysis bullosa with pyloric atresia. *Journal of Investigative Dermatology*, **2008**, 128, 2815-9	4.3	21
121	Persistent unilateral orbital and eyelid oedema as a manifestation of Melkersson-Rosenthal syndrome. *Journal of the European Academy of Dermatology and Venereology*, **2005**, 19, 107-11	4.6	21
120	Hereditary palmoplantar keratodermas. Part II: syndromic palmoplantar keratodermas - Diagnostic algorithm and principles of therapy. *Journal of the European Academy of Dermatology and Venereology*, **2018**, 32, 899-925	4.6	20
119	The evaluation of family impact of recessive dystrophic epidermolysis bullosa using the Italian version of the Family Dermatology Life Quality Index. *Journal of the European Academy of Dermatology and Venereology*, **2013**, 27, 1151-5	4.6	20
118	Sequential intramolecular epitope spreading of humoral responses to human BPAG2 in a transgenic model. *Journal of Investigative Dermatology*, **2010**, 130, 1040-7	4.3	20

117	Denaturing HPLC-based approach for detection of COL7A1 gene mutations causing dystrophic epidermolysis bullosa. *Biochemical and Biophysical Research Communications*, **2005**, 338, 1391-401	3.4	20
116	Trisomic rescue causing reduction to homozygosity for a novel ABCA12 mutation in harlequin ichthyosis. *Clinical Genetics*, **2009**, 76, 392-7	4	19
115	Inhibition of endothelial cell migration and angiogenesis by a vascular endothelial growth factor receptor-1 derived peptide. *European Journal of Cancer*, **2008**, 44, 1914-21	7.5	19
114	Intracellular degradation of beta4 integrin in lethal junctional epidermolysis bullosa with pyloric atresia. *British Journal of Dermatology*, **2004**, 151, 796-802	4	19
113	Different phenotypes in recessive dystrophic epidermolysis bullosa patients sharing the same mutation in compound heterozygosity with two novel mutations in the type VII collagen gene. *British Journal of Dermatology*, **2002**, 147, 450-7	4	19
112	Two novel recessive mutations in KRT14 identified in a cohort of 21 Spanish families with epidermolysis bullosa simplex. *British Journal of Dermatology*, **2011**, 165, 683-92	4	18
111	Circumscribed palmo-plantar hypokeratosis: a disease of desquamation? Immunohistological study of five cases and literature review. *Journal of the European Academy of Dermatology and Venereology*, **2011**, 25, 296-301	4.6	18
110	Glycated fibroblast growth factor-2 is quickly produced in vitro upon low-millimolar glucose treatment and detected in vivo in diabetic mice. *Molecular Endocrinology*, **2006**, 20, 2806-18		18
109	Gene correction of integrin beta4-dependent pyloric atresia-junctional epidermolysis bullosa keratinocytes establishes a role for beta4 tyrosines 1422 and 1440 in hemidesmosome assembly. *Journal of Biological Chemistry*, **2001**, 276, 41336-42	5.4	18
108	Fibroblastic rheumatism: a case without rheumatological symptoms. *Acta Dermato-Venereologica*, **2002**, 82, 200-3	2.2	18
107	Two families confirm Schöpf-Schulz-Passarge syndrome as a discrete entity within the WNT10A phenotypic spectrum. *Clinical Genetics*, **2011**, 79, 92-5	4	17
106	Herlitz junctional epidermolysis bullosa: laminin-5 mutational profile and carrier frequency in the Italian population. *British Journal of Dermatology*, **2008**, 158, 38-44	4	17
105	Association of piebaldism and neurofibromatosis type 1 in a girl. *Pediatric Dermatology*, **2001**, 18, 490-3	1.9	17
104	X-linked ichthyosis: Clinical and molecular findings in 35 Italian patients. *Experimental Dermatology*, **2019**, 28, 1156-1163	4	17
103	Induction of senescence pathways in Kindler syndrome primary keratinocytes. *British Journal of Dermatology*, **2013**, 168, 1019-26	4	16
102	European Guidelines (S3) on diagnosis and management of mucous membrane pemphigoid, initiated by the European Academy of Dermatology and Venereology - Part II. *Journal of the European Academy of Dermatology and Venereology*, **2021**, 35, 1926-1948	4.6	16
101	Epidermolysis Bullosa Simplex with KLHL24 Mutations Is Associated with Dilated Cardiomyopathy. *Journal of Investigative Dermatology*, **2019**, 139, 244-249	4.3	16
100	A founder synonymous COL7A1 mutation in three Danish families with dominant dystrophic epidermolysis bullosa pruriginosa identifies exonic regulatory sequences required for exon 87 splicing. *British Journal of Dermatology*, **2011**, 165, 678-82	4	15

99	Molecular testing in epidermolysis bullosa. *Dermatologic Clinics*, **2010**, 28, 223-9, vii-viii	4.2	15
98	Novel and recurrent mutations in the integrin beta 4 subunit gene causing lethal junctional epidermolysis bullosa with pyloric atresia. *Experimental Dermatology*, **2003**, 12, 716-20	4	15
97	A missense mutation (G1506E) in the adhesion G domain of laminin-5 causes mild junctional epidermolysis bullosa. *Biochemical and Biophysical Research Communications*, **2003**, 309, 96-103	3.4	15
96	Novel transglutaminase 1 mutations in patients affected by lamellar ichthyosis. *Cell Death and Disease*, **2012**, 3, e416	9.8	14
95	Novel and recurrent ALDH3A2 mutations in Italian patients with Sjögren-Larsson syndrome. *Journal of Human Genetics*, **2007**, 52, 865-870	4.3	14
94	Immunity to extracellular matrix antigens is associated with ultrastructural alterations of the stroma and stratified epithelium basement membrane in the skin of Hashimotos thyroiditis patients. *International Journal of Immunopathology and Pharmacology*, **2006**, 19, 661-74	3	14
93	Early immunopathological diagnosis of ichthyosis with confetti in two sporadic cases with new mutations in keratin 10. *Acta Dermato-Venereologica*, **2014**, 94, 579-82	2.2	13
92	Kindlin-1 mutant zebrafish as an in vivo model system to study adhesion mechanisms in the epidermis. *Journal of Investigative Dermatology*, **2013**, 133, 2180-90	4.3	13
91	Systematized organoid epidermal nevus with eccrine differentiation, multiple facial and oral congenital scars, gingival synechiae, and blepharophimosis: a novel epidermal nevus syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 25-31	2.5	13
90	180-kDa bullous pemphigoid antigen defective generalized atrophic benign epidermolysis bullosa: report of four cases with an unusually mild phenotype. *British Journal of Dermatology*, **1998**, 138, 859-66	4	13
89	A case of neonatal linear IgA bullous dermatosis with severe eye involvement. *Acta Dermato-Venereologica*, **2015**, 95, 1015-7	2.2	12
88	Reference genes for gene expression analysis in proliferating and differentiating human keratinocytes. *Experimental Dermatology*, **2015**, 24, 314-6	4	12
87	Endothelial cell adhesion to soluble vascular endothelial growth factor receptor-1 triggers a cell dynamic and angiogenic phenotype. *FASEB Journal*, **2014**, 28, 692-704	0.9	12
86	Novel physiological RECQL4 alternative transcript disclosed by molecular characterisation of Rothmund-Thomson Syndrome sibs with mild phenotype. *European Journal of Human Genetics*, **2014**, 22, 1298-304	5.3	12
85	Lethal autosomal recessive epidermolytic ichthyosis due to a novel donor splice-site mutation in KRT10. *British Journal of Dermatology*, **2010**, 162, 1384-7	4	12
84	A new SPINK5 donor splice site mutation in siblings with Netherton syndrome. *Acta Dermato-Venereologica*, **2010**, 90, 95-6	2.2	12
83	A synonymous mutation in SPINK5 exon 11 causes Netherton syndrome by altering exonic splicing regulatory elements. *Journal of Human Genetics*, **2012**, 57, 311-5	4.3	12
82	Impaired keratinocyte proliferative and clonogenic potential in transgenic mice overexpressing 14-3-3 in the epidermis. *Journal of Investigative Dermatology*, **2011**, 131, 1821-9	4.3	11

81	COL7A1 Recessive mutations in two siblings with distinct subtypes of dystrophic epidermolysis bullosa: pruriginosa versus nails only. *Dermatology*, **2011**, 222, 10-4	4.4	11
80	Phacomatosis cesioflammea with unilateral lipohypoplasia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 492-5	2.5	11
79	New type of epidermal nevus syndrome. *Dermatology*, **2000**, 201, 51-3	4.4	11
78	Notch-ing up knowledge on molecular mechanisms of skin fibrosis: focus on the multifaceted Notch signalling pathway. *Journal of Biomedical Science*, **2021**, 28, 36	13.3	11
77	Cockayne Syndrome Type A Protein Protects Primary Human Keratinocytes from Senescence. *Journal of Investigative Dermatology*, **2019**, 139, 38-50	4.3	11
76	Betapapillomavirus in multiple non-melanoma skin cancers of Netherton syndrome: Case report and published work review. *Journal of Dermatology*, **2015**, 42, 786-94	1.6	10
75	A novel frameshift mutation in the KIND1 gene in Turkish siblings with Kindler syndrome. *British Journal of Dermatology*, **2008**, 158, 1375-7	4	10
74	Biallelic somatic inactivation of the mismatch repair gene MLH1 in a primary skin melanoma. *Genes Chromosomes and Cancer*, **2003**, 37, 165-75	5	10
73	Molecular characterization of 11 Italian patients with Darier disease. *European Journal of Dermatology*, **2011**, 21, 334-8	0.8	10
72	European guidelines (S3) on diagnosis and management of mucous membrane pemphigoid, initiated by the European Academy of Dermatology and Venereology - Part I. *Journal of the European Academy of Dermatology and Venereology*, **2021**, 35, 1750-1764	4.6	10
71	MicroRNA-145-5p regulates fibrotic features of recessive dystrophic epidermolysis bullosa skin fibroblasts. *British Journal of Dermatology*, **2019**, 181, 1017-1027	4	9
70	Kindler syndrome with severe mucosal involvement in a large Palestinian pedigree. *European Journal of Dermatology*, **2015**, 25, 14-9	0.8	9
69	Assessment of the risk and characterization of non-melanoma skin cancer in Kindler syndrome: study of a series of 91 patients. *Orphanet Journal of Rare Diseases*, **2019**, 14, 183	4.2	9
68	Lethal Netherton syndrome due to homozygous p.Arg371X mutation in SPINK5. *Pediatric Dermatology*, **2013**, 30, e65-7	1.9	9
67	FERMT1 promoter mutations in patients with Kindler syndrome. *Clinical Genetics*, **2015**, 88, 248-54	4	9
66	Sensitivity of different assays for the serological diagnosis of epidermolysis bullosa acquisita: analysis of a cohort of 24 Italian patients. *Journal of the European Academy of Dermatology and Venereology*, **2014**, 28, 483-90	4.6	9
65	Long-term follow-up of a spontaneously improving patient with junctional epidermolysis bullosa associated with ITGB4 c.3977-19T>A splicing mutation. *Acta Dermato-Venereologica*, **2013**, 93, 116-8	2.2	9
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