Giovanna Zambruno

List of Publications by Year in Descending Order

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	Prominent Follicular Keratosis in Multiple Intestinal Atresia with Combined Immune Deficiency Caused by a TTC7A Homozygous Mutation. *Genes*, **2022**, 13, 821	4.2	
223	A multicenter study on quality of life of the "greater patient" in congenital ichthyoses. *Orphanet Journal of Rare Diseases*, **2021**, 16, 440	4.2	0
222	A Novel Phenotype of Junctional Epidermolysis Bullosa with Transient Skin Fragility and Predominant Ocular Involvement Responsive to Human Amniotic Membrane Eyedrops. *Genes*, **2021**, 12,	4.2	1
221	Ectodermal Dysplasia-Syndactyly Syndrome with Toe-Only Minimal Syndactyly Due to a Novel Mutation in NECTIN4: A Case Report and Literature Review. *Genes*, **2021**, 12,	4.2	0
220	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
219	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
218	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
217	Immunofluorescence mapping, electron microscopy and genetics in the diagnosis and sub-classification of inherited epidermolysis bullosa: a single-centre retrospective comparative study of 87 cases with long-term follow-up. *Journal of the European Academy of Dermatology and Venereology*, **2021**, 35, 1007-1016	4.6	6
216	Two Italian Patients with -Related Neuro-Ichthyosis: Expanding the Genotypic and Phenotypic Spectrum and Ultrastructural Characterization. *Genes*, **2021**, 12,	4.2	1
215	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	4.6	2
214	First Case of Epidermolytic Nevus and Novel Clinical and Genetic Findings in 26 Italian Patients with Keratinopathic Ichthyoses. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	4
213	Emergency management in epidermolysis bullosa: consensus clinical recommendations from the European reference network for rare skin diseases. *Orphanet Journal of Rare Diseases*, **2020**, 15, 142	4.2	3
212	Autosomal recessive epidermolysis bullosa simplex due to EXPH5 mutation: neonatal diagnosis of the first Italian case and literature review. *Journal of the European Academy of Dermatology and Venereology*, **2020**, 34, e694-e697	4.6	2
211	Multiple Skin Squamous Cell Carcinomas in Junctional Epidermolysis Bullosa Due to Altered Laminin-332 Function. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	1
210	Hoarse cry in a newborn with epidermolysis bullosa simplex, generalized severe. *Pediatric Dermatology*, **2020**, 37, 393-395	1.9	1
209	Self-improving dominant dystrophic epidermolysis bullosa: phenotypic variability associated with COL7A1 mutation p.Gly2037Glu. *European Journal of Dermatology*, **2020**, 30, 753-754	0.8	1
208	Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *British Journal of Dermatology*, **2020**, 183, 614-627	4	179

207	Women in Leadership and Scientific Development of the ESDR. *Journal of Investigative Dermatology* , **2020**, 140, S160-S162	4.3	
206	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
205	Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. *British Journal of Dermatology*, **2020**, 182, 574-592	4	45
204	Next Generation Sequencing Uncovers a Rare Case of X-linked Ichthyosis in an Adopted Girl Homozygous for a Novel Nonsense Mutation in the STS Gene. *Acta Dermato-Venereologica*, **2019**, 99, 828-830	2.2	2
203	Italian translation, cultural adaptation, and pilot testing of a questionnaire to assess family burden in inherited ichthyoses. *Italian Journal of Pediatrics*, **2019**, 45, 26	3.2	3
202	MicroRNA-145-5p regulates fibrotic features of recessive dystrophic epidermolysis bullosa skin fibroblasts. *British Journal of Dermatology*, **2019**, 181, 1017-1027	4	9
201	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
200	Palmoplantar Keratoderma and Woolly Hair Revealing Asymptomatic Arrhythmogenic Cardiomyopathy. *Acta Dermato-Venereologica*, **2019**, 99, 831-832	2.2	1
199	Identification of a Novel Non-desmoglein Autoantigen in Pemphigus Vulgaris. *Frontiers in Immunology*, **2019**, 10, 1391	8.4	2
198	Epidermolysis Bullosa-Associated Squamous Cell Carcinoma: From Pathogenesis to Therapeutic Perspectives. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	32
197	Epidermolysis Bullosa Simplex with KLHL24 Mutations Is Associated with Dilated Cardiomyopathy. *Journal of Investigative Dermatology*, **2019**, 139, 244-249	4.3	16
196	Decorin counteracts disease progression in mice with recessive dystrophic epidermolysis bullosa. *Matrix Biology*, **2019**, 81, 3-16	11.4	25
195	Teledermatology diagnosis of the first Italian patient affected with restrictive dermopathy due to ZMPSTE24 homozygous mutation. *Journal of the European Academy of Dermatology and Venereology*, **2019**, 33, e139-e140	4.6	2
194	Large International Validation of ABSIS and PDAI Pemphigus Severity Scores. *Journal of Investigative Dermatology*, **2019**, 139, 31-37	4.3	33
193	Cockayne Syndrome Type A Protein Protects Primary Human Keratinocytes from Senescence. *Journal of Investigative Dermatology*, **2019**, 139, 38-50	4.3	11
192	X-linked ichthyosis: Clinical and molecular findings in 35 Italian patients. *Experimental Dermatology*, **2019**, 28, 1156-1163	4	17
191	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
190	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

189	Local anesthesia in pediatric dermatologic surgery: Evaluation of a patient-centered approach. *Pediatric Dermatology*, **2018**, 35, 112-116	1.9	1
188	Lack of K140 immunoreactivity in junctional epidermolysis bullosa skin and keratinocytes associates with misfolded laminin epidermal growth factor-like motif 2 of the β short arm. *British Journal of Dermatology*, **2018**, 178, 1416-1422	4	7
187	Autosomal recessive epidermolysis bullosa simplex due to KRT14 mutation: two large Palestinian families and literature review. *Journal of the European Academy of Dermatology and Venereology*, **2018**, 32, e149-e151	4.6	2
186	Novel PNPLA1 mutations in two Italian siblings with autosomal recessive congenital ichthyosis. *Journal of the European Academy of Dermatology and Venereology*, **2018**, 32, e110-e112	4.6	5
185	Childhood epidermolysis bullosa acquisita during squaric acid dibutyl ester immunotherapy for alopecia areata. *British Journal of Dermatology*, **2017**, 176, 491-494	4	7
184	Stromal microenvironment in type VII collagen-deficient skin: The ground for squamous cell carcinoma development. *Matrix Biology*, **2017**, 63, 1-10	11.4	58
183	IgA tracheobronchial deposits underlie respiratory compromise in neonatal linear IgA bullous dermatosis. *Journal of the European Academy of Dermatology and Venereology*, **2017**, 31, e333-e335	4.6	2
182	Pathomechanisms of Altered Wound Healing in Recessive Dystrophic Epidermolysis Bullosa. *American Journal of Pathology*, **2017**, 187, 1445-1453	5.8	34
181	Severe osteoarticular involvement in isotretinoin-triggered acne fulminans: two cases successfully treated with anakinra. *Journal of the European Academy of Dermatology and Venereology*, **2017**, 31, e277-e279	4.6	6
180	Lipoid Proteinosis: A Previously Unrecognized Mutation and Therapeutic Response to Acitretin. *Acta Dermato-Venereologica*, **2017**, 97, 1249-1251	2.2	1
179	Monoallelic Mutations in the Translation Initiation Codon of KLHL24 Cause Skin Fragility. *American Journal of Human Genetics*, **2016**, 99, 1395-1404	11	51
178	A compound synonymous mutation c.474G>A with p.Arg578X mutation in SPINK5 causes splicing disorder and mild phenotype in Netherton syndrome. *Experimental Dermatology*, **2016**, 25, 568-70	4	4
177	Frequent Occurrence of Aplasia Cutis Congenita in Bullous Dermolysis of the Newborn. *Acta Dermato-Venereologica*, **2016**, 96, 784-7	2.2	4
176	Structural Defects of Laminin β N-terminus Underlie Junctional Epidermolysis Bullosa with Altered Granulation Tissue Response. *Acta Dermato-Venereologica*, **2016**, 96, 954-958	2.2	4
175	Pemphigoid Gestationis Complicating an Egg Donation Pregnancy. *Acta Dermato-Venereologica*, **2016**, 96, 695-6	2.2	2
174	Paraneoplastic Epidermolysis Bullosa Acquisita Associated with Thyroid Carcinoma. *Acta Dermato-Venereologica*, **2016**, 96, 414-5	2.2	2
173	Acral skin atrophy in an infant: an early clue to Kindler syndrome diagnosis. *Journal of the European Academy of Dermatology and Venereology*, **2016**, 30, 1046-9	4.6	5
172	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

171	Clonal analysis of B-cell response in pemphigus course: toward more effective therapies. *Journal of Investigative Dermatology*, **2015**, 135, 651-654	4.3	2
170	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
169	Interleukin-22 Promotes Wound Repair in Diabetes by Improving Keratinocyte Pro-Healing Functions. *Journal of Investigative Dermatology*, **2015**, 135, 2862-2870	4.3	50
168	Kindler syndrome with severe mucosal involvement in a large Palestinian pedigree. *European Journal of Dermatology*, **2015**, 25, 14-9	0.8	9
167	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
166	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
165	Recessive bullous dermolysis of the newborn in preterm siblings with a missense mutation in type VII collagen. *Pediatric Dermatology*, **2015**, 32, e42-7	1.9	5
164	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
163	Ichthyosis Linearis Circumflexa as the Only Clinical Manifestation of Netherton Syndrome. *Acta Dermato-Venereologica*, **2015**, 95, 720-4	2.2	4
162	Ichthyosis with confetti: clinics, molecular genetics and management. *Orphanet Journal of Rare Diseases*, **2015**, 10, 115	4.2	21
161	FERMT1 promoter mutations in patients with Kindler syndrome. *Clinical Genetics*, **2015**, 88, 248-54	4	9
160	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
159	A case of neonatal linear IgA bullous dermatosis with severe eye involvement. *Acta Dermato-Venereologica*, **2015**, 95, 1015-7	2.2	12
158	The E3 ligase Itch knockout mice show hyperproliferation and wound healing alteration. *FEBS Journal*, **2015**, 282, 4435-49	5.7	8
157	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
156	Whole-exome sequencing in patients with ichthyosis reveals modifiers associated with increased IgE levels and allergic sensitizations. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 280-3	11.5	8
155	Reference genes for gene expression analysis in proliferating and differentiating human keratinocytes. *Experimental Dermatology*, **2015**, 24, 314-6	4	12
154	Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. *Journal of the American Academy of Dermatology*, **2014**, 70, 1103-26	4.5	596

153	Multicentre consensus recommendations for skin care in inherited epidermolysis bullosa. *Orphanet Journal of Rare Diseases*, **2014**, 9, 76	4.2	76
152	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
151	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
150	T-lymphocytes are directly involved in the clinical expression of migratory circinate erythema in epidermolysis bullosa simplex patients. *Acta Dermato-Venereologica*, **2014**, 94, 307-11	2.2	7
149	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
148	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
147	A truncating mutation in the laminin-332 chain highlights the role of the LG45 proteolytic domain in regulating keratinocyte adhesion and migration. *British Journal of Dermatology*, **2014**, 170, 1056-64	4	8
146	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
145	Monozygotic twins discordant for recessive dystrophic epidermolysis bullosa phenotype highlight the role of TGF- signalling in modifying disease severity. *Human Molecular Genetics*, **2014**, 23, 3907-22	5.6	61
144	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
143	Lethal Netherton syndrome due to homozygous p.Arg371X mutation in SPINK5. *Pediatric Dermatology*, **2013**, 30, e65-7	1.9	9
142	Skin equivalents: a tool for the discovery and validation of pharmacodynamic biomarkers. *Cancer Investigation*, **2013**, 31, 60-6	2.1	1
141	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
140	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
139	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
138	Induction of senescence pathways in Kindler syndrome primary keratinocytes. *British Journal of Dermatology*, **2013**, 168, 1019-26	4	16
137	Kindlin-1 regulates integrin dynamics and adhesion turnover. *PLoS ONE*, **2013**, 8, e65341	3.7	22
136	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

135	Bullous pemphigoid: from the clinic to the bench. *Clinics in Dermatology*, **2012**, 30, 3-16	3	96
134	Prevalence of collagen VII-specific autoantibodies in patients with autoimmune and inflammatory diseases. *BMC Immunology*, **2012**, 13, 16	3.7	27
133	The atypical retinoid E-3-(3PAdamantan-1-yl-4Pmethoxybiphenyl-4-yl)-2-propenoic acid (ST1898) displays comedolytic activity in the rhino mouse model. *European Journal of Dermatology*, **2012**, 22, 505-11	9.8	3
132	Endemic pemphigus foliaceus: towards understanding autoimmune mechanisms of disease development. *Journal of Investigative Dermatology*, **2012**, 132, 2499-502	4.3	8
131	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
130	Novel transglutaminase 1 mutations in patients affected by lamellar ichthyosis. *Cell Death and Disease*, **2012**, 3, e416	9.8	14
129	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
128	Pemphigus autoantibodies generated through somatic mutations target the desmoglein-3 cis-interface. *Journal of Clinical Investigation*, **2012**, 122, 3781-90	15.9	112
127	Two families confirm Schöpf-Schulz-Passarge syndrome as a discrete entity within the WNT10A phenotypic spectrum. *Clinical Genetics*, **2011**, 79, 92-5	4	17
126	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
125	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
124	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
123	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
122	AXIN2 germline mutations are rare in familial melanoma. *Genes Chromosomes and Cancer*, **2011**, 50, 370-3	3	6
121	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
120	Kindler syndrome: extension of FERMT1 mutational spectrum and natural history. *Human Mutation*, **2011**, 32, 1204-12	4.7	82
119	Impaired keratinocyte proliferative and clonogenic potential in transgenic mice overexpressing 14-3-3in the epidermis. *Journal of Investigative Dermatology*, **2011**, 131, 1821-9	4.3	11
118	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

117	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
116	Sensitivity of immunofluorescence studies vs enzyme-linked immunosorbent assay for diagnosis of bullous pemphigoid. *Archives of Dermatology*, **2011**, 147, 1454-6; author reply 1456		3
115	A recurrent nonsense mutation occurring as a de novo event in a patient with recessive dystrophic epidermolysis bullosa. *Dermatology*, **2011**, 223, 219-21	4.4	6
114	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
113	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
112	Sirtinol treatment reduces inflammation in human dermal microvascular endothelial cells. *PLoS ONE*, **2011**, 6, e24307	3.7	49
111	Molecular characterization of 11 Italian patients with Darier disease. *European Journal of Dermatology*, **2011**, 21, 334-8	0.8	10
110	Bullous Pemphigoid: Clinical Features, Diagnostic Markers, and Immunopathogenic Mechanisms **2011**, 65-95		
109	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
108	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
107	TSH receptor and thyroid-specific gene expression in human skin. *Journal of Investigative Dermatology*, **2010**, 130, 93-101	4.3	75
106	A new SPINK5 donor splice site mutation in siblings with Netherton syndrome. *Acta Dermato-Venereologica*, **2010**, 90, 95-6	2.2	12
105	Family burden in epidermolysis bullosa is high independent of disease type/subtype. *Acta Dermato-Venereologica*, **2010**, 90, 607-11	2.2	25
104	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
103	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
102	Epidermolysis bullosa care in Italy. *Dermatologic Clinics*, **2010**, 28, 407-9, xiv-xv	4.2	7
101	Mutation mechanisms. *Dermatologic Clinics*, **2010**, 28, 17-22	4.2	4
100	Molecular testing in epidermolysis bullosa. *Dermatologic Clinics*, **2010**, 28, 223-9, vii-viii	4.2	15

99	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
98	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
97	Genotype-phenotype relationships in trichothiodystrophy patients with novel splicing mutations in the XPD gene. *Human Mutation*, **2009**, 30, 438-45	4.7	21
96	Branch point and donor splice-site COL7A1 mutations in mild recessive dystrophic epidermolysis bullosa. *British Journal of Dermatology*, **2009**, 161, 464-7	4	3
95	Quality of life in patients with epidermolysis bullosa. *British Journal of Dermatology*, **2009**, 161, 869-77	4	61
94	Mutations in PYCR1 cause cutis laxa with progeroid features. *Nature Genetics*, **2009**, 41, 1016-21	36.3	178
93	Trisomic rescue causing reduction to homozygosity for a novel ABCA12 mutation in harlequin ichthyosis. *Clinical Genetics*, **2009**, 76, 392-7	4	19
92	Clinical and genetic heterogeneity in keratosis follicularis spinulosa decalvans. *European Journal of Medical Genetics*, **2009**, 52, 53-8	2.6	23
91	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
90	Rituximab immunotherapy in pemphigus: therapeutic effects beyond B-cell depletion. *Journal of Investigative Dermatology*, **2008**, 128, 2745-7	4.3	34
89	A novel frameshift mutation in the KIND1 gene in Turkish siblings with Kindler syndrome. *British Journal of Dermatology*, **2008**, 158, 1375-7	4	10
88	Multicenter prospective study of the humoral autoimmune response in bullous pemphigoid. *Clinical Immunology*, **2008**, 128, 415-26	9	141
87	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
86	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
85	A proangiogenic peptide derived from vascular endothelial growth factor receptor-1 acts through alpha5beta1 integrin. *Blood*, **2008**, 111, 3479-88	2.2	25
84	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
83	Concomitant activation of Wnt pathway and loss of mismatch repair function in human melanoma. *Genes Chromosomes and Cancer*, **2008**, 47, 614-24	5	26
82	Phacomatosis cesioflammea with unilateral lipohypoplasia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 492-5	2.5	11

81	Association of segmental neurofibromatosis 1 and oculo-auriculo-vertebral spectrum in a 24-year-old female. *European Journal of Dermatology*, **2008**, 18, 22-5	0.8	6
80	Schöf-Schulz-Passarge syndrome: further delineation of the phenotype and genetic considerations. *Acta Dermato-Venereologica*, **2008**, 88, 607-12	2.2	8
79	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
78	The role of CSA in the response to oxidative DNA damage in human cells. *Oncogene*, **2007**, 26, 4336-43	9.2	126
77	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
76	Delayed diagnosis of dyskeratosis congenita in a 40-year-old woman with multiple head and neck squamous cell carcinomas. *British Journal of Dermatology*, **2007**, 156, 406-8	4	5
75	Oral pemphigoid autoantibodies preferentially target BP180 ectodomain. *Clinical Immunology*, **2007**, 122, 207-13	9	42
74	Novel and recurrent ALDH3A2 mutations in Italian patients with Sjögren-Larsson syndrome. *Journal of Human Genetics*, **2007**, 52, 865-870	4.3	14
73	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
72	Distinct vascular endothelial growth factor signals for lymphatic vessel enlargement and sprouting. *Journal of Experimental Medicine*, **2007**, 204, 1431-40	16.6	137
71	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
70	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
69	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
68	Increased melanoma growth and metastasis spreading in mice overexpressing placenta growth factor. *American Journal of Pathology*, **2006**, 169, 643-54	5.8	83
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