Daniele Castiglia

List of Publications by Year
in descending order

113	2,592	172457	254184
papers	citations	29 h-index	43 g-index

114	114	114	2997
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prominent Follicular Keratosis in Multiple Intestinal Atresia with Combined Immune Deficiency Caused by a TTC7A Homozygous Mutation. Genes, 2022, 13, 821.	2.4	1
2	Immunofluorescence mapping, electron microscopy and genetics in the diagnosis and subâ€classification of inherited epidermolysis bullosa: a singleâ€centre retrospective comparative study of 87 cases with longâ€term followâ€up. Journal of the European Academy of Dermatology and Venereology, 2021, 35, 1007-1016.	2.4	10
3	The Burden of Autosomal Recessive Congenital Ichthyoses on Patients and their Families: An Italian Multicentre Study. Acta Dermato-Venereologica, 2021, 101, adv00477.	1.3	10
4	A Novel Phenotype of Junctional Epidermolysis Bullosa with Transient Skin Fragility and Predominant Ocular Involvement Responsive to Human Amniotic Membrane Eyedrops. Genes, 2021, 12, 716.	2.4	5
5	Notch-ing up knowledge on molecular mechanisms of skin fibrosis: focus on the multifaceted Notch signalling pathway. Journal of Biomedical Science, 2021, 28, 36.	7.0	33
6	RSPO1-mutated fibroblasts from non-tumoural areas of palmoplantar keratoderma display a cancer-associated phenotype. European Journal of Dermatology, 2021, 31, 342-350.	0.6	2
7	Development of bullous pemphigoid in junctional epidermolysis bullosa. Journal of the European Academy of Dermatology and Venereology, 2020, 34, e146-e148.	2.4	4
8	First Case of KRT2 Epidermolytic Nevus and Novel Clinical and Genetic Findings in 26 Italian Patients with Keratinopathic Ichthyoses. International Journal of Molecular Sciences, 2020, 21, 7707.	4.1	10
9	Multiple Skin Squamous Cell Carcinomas in Junctional Epidermolysis Bullosa Due to Altered Laminin-332 Function. International Journal of Molecular Sciences, 2020, 21, 1426.	4.1	3
10	A previously unrecognized Ankyrinâ€1 mutation associated with Hereditary Spherocytosis in an Italian family. European Journal of Haematology, 2019, 103, 523-526.	2.2	3
11	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.	2.7	16
12	Palmoplantar Keratoderma and Woolly Hair Revealing Asymptomatic Arrhythmogenic Cardiomyopathy. Acta Dermato-Venereologica, 2019, 99, 831-832.	1.3	4
13	302 Molecular and mutational signatures of squamous cell carcinomas in epidermolysis bullosa. Journal of Investigative Dermatology, 2019, 139, S266.	0.7	1
14	MicroRNAâ€145â€5p regulates fibrotic features of recessive dystrophic epidermolysis bullosa skin fibroblasts. British Journal of Dermatology, 2019, 181, 1017-1027.	1.5	19
15	Epidermolysis Bullosa-Associated Squamous Cell Carcinoma: From Pathogenesis to Therapeutic Perspectives. International Journal of Molecular Sciences, 2019, 20, 5707.	4.1	72
16	miRâ€145â€5p profibrotic activity in RDEB. British Journal of Dermatology, 2019, 181, e121.	1.5	0
17	RDEB ä¸š„ miRâ€145â€5p ä¿ƒçº»åŒ–æ´»æ€§. British Journal of Dermatology, 2019, 181, e133.	1.5	0
18	Epidermolysis Bullosa Simplex with KLHL24 Mutations Is Associated with Dilated Cardiomyopathy. Journal of Investigative Dermatology, 2019, 139, 244-249.	0.7	23

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19	Phenotypic Features of Epidermolysis Bullosa Simplex due to KLHL24 Mutations in 3 Italian Cases. Acta Dermato-Venereologica, 2019, 99, 238-239.	1.3	7
20	Recessive mutations in the neuronal isoforms of DST , encoding dystonin, lead to abnormal actin cytoskeleton organization and HSAN type VI. Human Mutation, 2019, 40, 106-114.	2.5	30
21	Decorin counteracts disease progression in mice with recessive dystrophic epidermolysis bullosa. Matrix Biology, 2019, 81, 3-16.	3.6	38
22	Hereditary palmoplantar keratodermas. Part I. Nonâ€syndromic palmoplantar keratodermas: classification, clinical and genetic features. Journal of the European Academy of Dermatology and Venereology, 2018, 32, 704-719.	2.4	47
23	Hereditary palmoplantar keratodermas. Part <scp>II</scp>: syndromic palmoplantar keratodermas â€“ Diagnostic algorithm and principles of therapy. Journal of the European Academy of Dermatology and Venereology, 2018, 32, 899-925.	2.4	34
24	Lack of K140 immunoreactivity in junctional epidermolysis bullosa skin and keratinocytes associates with misfolded laminin epidermal growth factor-like motif 2 of the Î²3 short arm. British Journal of Dermatology, 2018, 178, 1416-1422.	1.5	7
25	Autosomal recessive epidermolysis bullosa simplex due to <scp>KRT</scp>14 mutation: two large Palestinian families and literature review. Journal of the European Academy of Dermatology and Venereology, 2018, 32, e149-e151.	2.4	3
26	Microprocessor-dependent processing of splice site overlapping microRNA exons does not result in changes in alternative splicing. Rna, 2018, 24, 1158-1171.	3.5	12
27	A Postzygotic ATP2A2 Novel Mutation Identified by Next-generation Sequencing in Mosaic Darier Disease. Acta Dermato-Venereologica, 2018, 99, 115-116.	1.3	4
28	Ichthyosis Prematurity Syndrome due to a Novel SLC27A4 Homozygous Mutation in an Italian Patient. Acta Dermato-Venereologica, 2018, 98, 803-804.	1.3	7
29	Childhood epidermolysis bullosa acquisita during squaric acid dibutyl ester immunotherapy for alopecia areata. British Journal of Dermatology, 2017, 176, 491-494.	1.5	11
30	Stromal microenvironment in type VII collagen-deficient skin: The ground for squamous cell carcinoma development. Matrix Biology, 2017, 63, 1-10.	3.6	81
31	Pathomechanisms of Altered Wound Healing in Recessive Dystrophic Epidermolysis Bullosa. American Journal of Pathology, 2017, 187, 1445-1453.	3.8	56
32	Lipoid Proteinosis: A Previously Unrecognized Mutation and Therapeutic Response to Acitretin. Acta Dermato-Venereologica, 2017, 97, 1249-1251.	1.3	2
33	Accelerated features of senescence in cultured type 2 diabetic skin fibroblasts. European Journal of Dermatology, 2017, 27, 408-410.	0.6	2
34	FOXM1 regulates proliferation, senescence and oxidative stress in keratinocytes and cancer cells. Aging, 2016, 8, 1384-1397.	3.1	57
35	A uniqueLAMB3splice-site mutation with founder effect from the Balkans causes lethal epidermolysis bullosa in several European countries. British Journal of Dermatology, 2016, 175, 721-727.	1.5	12
36	Acral skin atrophy in an infant: an early clue to Kindler syndrome diagnosis. Journal of the European Academy of Dermatology and Venereology, 2016, 30, 1046-1049.	2.4	6

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37	Monoallelic Mutations in the Translation Initiation Codon of KLHL24 Cause Skin Fragility. American Journal of Human Genetics, 2016, 99, 1395-1404.	6.2	71
38	Genetic basis of dominant dystrophic epidermolysis bullosa in tunisian families and coâ€•occurrence of dominant and recessive mutations. Journal of the European Academy of Dermatology and Venereology, 2016, 30, 155-157.	2.4	2
39	A compound synonymous mutation c.474G>A with p.Arg578X mutation in <scp>SPINK</scp>5 causes splicing disorder and mild phenotype in Netherton syndrome. Experimental Dermatology, 2016, 25, 568-570.	2.9	6
40	Ichthyosis with confetti: clinics, molecular genetics and management. Orphanet Journal of Rare Diseases, 2015, 10, 115.	2.7	30
41	<scp>FERMT1</scp> promoter mutations in patients with Kindler syndrome. Clinical Genetics, 2015, 88, 248-254.	2.0	11
42	The Increase in Maternal Expression of axin1 and axin2 Contribute to the Zebrafish Mutant Ichabod Ventralized Phenotype. Journal of Cellular Biochemistry, 2015, 116, 418-430.	2.6	7
43	Betapapillomavirus in multiple nonâ€•melanoma skin cancers of Netherton syndrome: Case report and published work review. Journal of Dermatology, 2015, 42, 786-794.	1.2	15
44	Kindler syndrome with severe mucosal involvement in a large Palestinian pedigree. European Journal of Dermatology, 2015, 25, 14-19.	0.6	11
45	Recessive Bullous Dermolysis of the Newborn in Preterm Siblings with a Missense Mutation in Type <scp>VII</scp> Collagen. Pediatric Dermatology, 2015, 32, e42-7.	0.9	8
46	Exon-Specific U1s Correct SPINK5Exon 11 Skipping Caused by a Synonymous Substitution that Affects a Bifunctional Splicing Regulatory Element. Human Mutation, 2015, 36, 504-512.	2.5	33
47	Novel physiological RECQL4 alternative transcript disclosed by molecular characterisation of Rothmundâ€“Thomson Syndrome sibs with mild phenotype. European Journal of Human Genetics, 2014, 22, 1298-1304.	2.8	13
48	T-lymphocytes are Directly Involved in the Clinical Expression of Migratory Circinate Erythema in Epidermolysis Bullosa Simplex Patients. Acta Dermato-Venereologica, 2014, 94, 307-311.	1.3	12
49	Early Immunopathological Diagnosis of Ichthyosis with Confetti in Two Sporadic Cases with New Mutations in Keratin 10. Acta Dermato-Venereologica, 2014, 94, 579-582.	1.3	17
50	A truncating mutation in the laminin-332Î± chain highlights the role of the LG45 proteolytic domain in regulating keratinocyte adhesion and migration. British Journal of Dermatology, 2014, 170, 1056-1064.	1.5	11
51	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.	0.7	33
52	Monozygotic twins discordant for recessive dystrophic epidermolysis bullosa phenotype highlight the role of TGF-Î² signalling in modifying disease severity. Human Molecular Genetics, 2014, 23, 3907-3922.	2.9	88
53	Mutational founder effect in recessive dystrophic epidermolysis bullosa families from Southern Tunisia. Archives of Dermatological Research, 2014, 306, 405-411.	1.9	5
54	Lethal Netherton Syndrome Due to Homozygous p.<scp>A</scp>rg371<scp>X</scp> Mutation in <scp>SPINK</scp>5. Pediatric Dermatology, 2013, 30, e65-7.	0.9	15

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55	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, 1151-1155.	2.4	26
56	PNA as a potential modulator of COL7A1 gene expression in dominant dystrophic epidermolysis bullosa: a physico-chemical study. Molecular BioSystems, 2013, 9, 3166.	2.9	9
57	Induction of senescence pathways in Kindler syndrome primary keratinocytes. British Journal of Dermatology, 2013, 168, 1019-1026.	1.5	18
58	A synonymous mutation in SPINK5 exon 11 causes Netherton syndrome by altering exonic splicing regulatory elements. Journal of Human Genetics, 2012, 57, 311-315.	2.3	12
59	Dystrophic epidermolysis bullosa pruriginosa with autoantibodies against collagen VII. European Journal of Dermatology, 2012, 22, 541-542.	0.6	4
60	Two families confirm SchÃ¶pf-Schulz-Passarge syndrome as a discrete entity within the WNT10A phenotypic spectrum. Clinical Genetics, 2011, 79, 92-95.	2.0	20
61	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, 678-682.	1.5	17
62	Two novel recessive mutations in KRT14 identified in a cohort of 21 Spanish families with epidermolysis bullosa simplex. British Journal of Dermatology, 2011, 165, 683-692.	1.5	24
63	AXIN2 germline mutations are rare in familial melanoma. Genes Chromosomes and Cancer, 2011, 50, 370-373.	2.8	8
64	The international dystrophic epidermolysis bullosa patient registry: An online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. Human Mutation, 2011, 32, 1100-1107.	2.5	74
65	Kindler syndrome: Extension of FERMT1 mutational spectrum and natural history. Human Mutation, 2011, 32, 1204-1212.	2.5	102
66	Impaired Keratinocyte Proliferative and Clonogenic Potential in Transgenic Mice Overexpressing 14-3-3Ïƒ in the Epidermis. Journal of Investigative Dermatology, 2011, 131, 1821-1829.	0.7	14
67	COL7A1 Recessive Mutations in Two Siblings with Distinct Subtypes of Dystrophic Epidermolysis Bullosa: Pruriginosa versus Nails Only. Dermatology, 2011, 222, 10-14.	2.1	16
68	Lethal autosomal recessive epidermolytic ichthyosis due to a novel donor splice-site mutation inKRT10. British Journal of Dermatology, 2010, 162, 1384-1387.	1.5	17
69	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, 155-161.	1.5	53
70	A New SPINK5 Donor Splice Site Mutation in Siblings with Netherton Syndrome. Acta Dermato-Venereologica, 2010, 90, 95-96.	1.3	14
71	Epidermolysis Bullosa Care in Italy. Dermatologic Clinics, 2010, 28, 407-409.	1.7	7
72	Mutation Mechanisms. Dermatologic Clinics, 2010, 28, 17-22.	1.7	6

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73	Mutational survey of recessive dystrophic epidermolysis bullosa in Tunisian families unveils a spectrum of private, ethnic specific and world wide recurrent mutations. Journal of Dermatological Science, 2010, 57, 144-146.	1.9	4
74	The cyclin-dependent kinase inhibitor PHA-848125 suppresses the in vitro growth of human melanomas sensitive or resistant to temozolomide, and shows synergistic effects in combination with this triazene compound. Pharmacological Research, 2010, 61, 437-448.	7.1	26
75	Molecular Testing in Epidermolysis Bullosa. Dermatologic Clinics, 2010, 28, 223-229.	1.7	27
76	NovelCTSCmutations in a patient with Papillon-LefÃ¨vre syndrome with recurrent pyoderma and minimal oral and palmoplantar involvement. British Journal of Dermatology, 2009, 160, 881-883.	1.5	13
77	Branch point and donor splice-siteCOL7A1mutations in mild recessive dystrophic epidermolysis bullosa. British Journal of Dermatology, 2009, 161, 464-467.	1.5	3
78	Quality of life in patients with epidermolysis bullosa. British Journal of Dermatology, 2009, 161, 869-877.	1.5	78
79	Trisomic rescue causing reduction to homozygosity for a novel ABCA12 mutation in harlequin ichthyosis. Clinical Genetics, 2009, 76, 392-397.	2.0	21
80	Bazexâ€“DuprÃ©â€“Christol syndrome: An ectodermal dysplasia with skin appendage neoplasms. European Journal of Medical Genetics, 2009, 52, 250-255.	1.3	22
81	Dystrophic epidermolysis bullosa phenotypes in a large consanguineous Tunisian family. Journal of Dermatological Science, 2009, 54, 114-120.	1.9	10
82	Concomitant activation of Wnt pathway and loss of mismatch repair function in human melanoma. Genes Chromosomes and Cancer, 2008, 47, 614-624.	2.8	34
83	Complete maternal isodisomy causing reduction to homozygosity for a novel LAMB3 mutation in Herlitz junctional epidermolysis bullosa. Journal of Dermatological Science, 2008, 51, 58-61.	1.9	15
84	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, 406-408.	1.5	9
85	Herlitz junctional epidermolysis bullosa: laminin-5 mutational profile and carrier frequency in the Italian population. British Journal of Dermatology, 2007, 158, 071004160508001-???.	1.5	24
86	Dystrophic epidermolysis bullosa pruriginosa in Italy: clinical and molecular characterization. Clinical Genetics, 2006, 70, 339-347.	2.0	33
87	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, 1776-1783.	0.7	54
88	Human Melanoma Metastases Express Functional CXCR4. Clinical Cancer Research, 2006, 12, 2427-2433.	7.0	114
89	Epidermolysis bullosa simplex with mottled pigmentation due to de novo P25L mutation in keratin 5 in an Italian patient. European Journal of Dermatology, 2006, 16, 620-2.	0.6	6
90	Analysis of candidate genes through a proteomics-based approach in primary cell lines from malignant melanomas and their metastases. Melanoma Research, 2005, 15, 235-244.	1.2	50

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91	Denaturing HPLC-based approach for detection of COL7A1 gene mutations causing dystrophic epidermolysis bullosa. Biochemical and Biophysical Research Communications, 2005, 338, 1391-1401.	2.1	23
92	BRAF Gene Is Somatically Mutated but Does Not Make a Major Contribution to Malignant Melanoma Susceptibility: The Italian Melanoma Intergroup Study. Journal of Clinical Oncology, 2004, 22, 286-292.	1.6	55
93	Laminin-5 Mutational Analysis in an Italian Cohort of Patients with Junctional Epidermolysis Bullosa. Journal of Investigative Dermatology, 2004, 123, 639-648.	0.7	33
94	Biallelic somatic inactivation of the mismatch repair gene MLH1 in a primary skin melanoma. Genes Chromosomes and Cancer, 2003, 37, 165-175.	2.8	16
95	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.	2.1	20
96	High-Frequency Microsatellite Instability is Associated with Defective DNA Mismatch Repair in Human Melanoma. Journal of Investigative Dermatology, 2002, 118, 79-86.	0.7	30
97	Genotypeâ€“Phenotype Correlation in Italian Patients with Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2002, 119, 1456-1462.	0.7	58
98	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, 182-187.	0.7	34
99	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, 731-739.	0.7	31
100	Pretibial dystrophic epidermolysis bullosa: a recessively inherited COL7A1 splice site mutation affecting procollagen VII processing. British Journal of Dermatology, 1999, 141, 833-839.	1.5	24
101	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, 758-764.	2.1	27
102	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, 744-750.	0.7	34
103	PIPPin, a Putative RNA-Binding Protein Specifically Expressed in the Rat Brain. Biochemical and Biophysical Research Communications, 1996, 218, 390-394.	2.1	36
104	Expression of synapsin I gene in primary cultures of differentiating rat cortical neurons. Neurochemical Research, 1995, 20, 239-243.	3.3	16
105	Posttranscriptional regulation of H1Â° and H3.3B histone genes in differentiating rat cortical neurons. Neurochemical Research, 1995, 20, 969-976.	3.3	25
106	H1Â° and H3.3B mRNA levels in developing rat brain. Neurochemical Research, 1994, 19, 1531-1537.	3.3	30
107	Cloning and analysis of cDNA for rat histone H1Â°. Nucleic Acids Research, 1993, 21, 1674-1674.	14.5	20
108	Qualitative differences in nuclear proteins correlate with neuronal terminal differentiation. Cellular and Molecular Neurobiology, 1992, 12, 33-43.	3.3	15

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109	Accumulation of different c-erbA transcripts during rat brain development and in cortical neurons cultured in a synthetic medium. Cellular and Molecular Neurobiology, 1992, 12, 259-272.	3.3	9
110	The dynamic properties of neuronal chromatin are modulated by triiodothyronine. Neurochemical Research, 1992, 17, 1049-1055.	3.3	10
111	Structural definition by antibody engineering of an idiotypic determinant. Protein Engineering, Design and Selection, 1990, 3, 531-539.	2.1	23
112	Autoantibody Idiotypy and Neonatal B Cell Repertoire. Viral Immunology, 1989, 2, 263-269.	1.3	0
113	Triiodothyronine-Induced Shortening of Chromatin Repeat Length in Neurons Cultured in a Chemically Denned Medium. Journal of Neurochemistry, 1987, 48, 1053-1059.	3.9	19