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	Human Melanoma Metastases Express Functional CXCR4. Clinical Cancer Research, 2006, 12, 2427-2433.	7.0	114
2	Kindler syndrome: Extension of FERMT1 mutational spectrum and natural history. Human Mutation, 2011, 32, 1204-1212.	2.5	102
3	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
4	Stromal microenvironment in type VII collagen-deficient skin: The ground for squamous cell carcinoma development. Matrix Biology, 2017, 63, 1-10.	3.6	81
5	Quality of life in patients with epidermolysis bullosa. British Journal of Dermatology, 2009, 161, 869-877.	1.5	78
6	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
7	Epidermolysis Bullosa-Associated Squamous Cell Carcinoma: From Pathogenesis to Therapeutic Perspectives. International Journal of Molecular Sciences, 2019, 20, 5707.	4.1	72
8	Monoallelic Mutations in the Translation Initiation Codon of KLHL24 Cause Skin Fragility. American Journal of Human Genetics, 2016, 99, 1395-1404.	6.2	71
9	Genotypeâ€“Phenotype Correlation in Italian Patients with Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2002, 119, 1456-1462.	0.7	58
10	FOXM1 regulates proliferation, senescence and oxidative stress in keratinocytes and cancer cells. Aging, 2016, 8, 1384-1397.	3.1	57
11	Pathomechanisms of Altered Wound Healing in Recessive Dystrophic Epidermolysis Bullosa. American Journal of Pathology, 2017, 187, 1445-1453.	3.8	56
12	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
13	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
14	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
15	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
16	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
17	Decorin counteracts disease progression in mice with recessive dystrophic epidermolysis bullosa. Matrix Biology, 2019, 81, 3-16.	3.6	38
18	PIPPin, a Putative RNA-Binding Protein Specifically Expressed in the Rat Brain. Biochemical and Biophysical Research Communications, 1996, 218, 390-394.	2.1	36

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19	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
20	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
21	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
22	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
23	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
24	Dystrophic epidermolysis bullosa pruriginosa in Italy: clinical and molecular characterization. Clinical Genetics, 2006, 70, 339-347.	2.0	33
25	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
26	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
27	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
28	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
29	H1Â° and H3.3B mRNA levels in developing rat brain. Neurochemical Research, 1994, 19, 1531-1537.	3.3	30
30	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
31	Ichthyosis with confetti: clinics, molecular genetics and management. Orphanet Journal of Rare Diseases, 2015, 10, 115.	2.7	30
32	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
33	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
34	Molecular Testing in Epidermolysis Bullosa. Dermatologic Clinics, 2010, 28, 223-229.	1.7	27
35	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
36	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

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37	Posttranscriptional regulation of H1Â° and H3.3B histone genes in differentiating rat cortical neurons. Neurochemical Research, 1995, 20, 969-976.	3.3	25
38	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
39	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
40	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
41	Structural definition by antibody engineering of an idiotypic determinant. Protein Engineering, Design and Selection, 1990, 3, 531-539.	2.1	23
42	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
43	Epidermolysis Bullosa Simplex with KLHL24 Mutations Is Associated with Dilated Cardiomyopathy. Journal of Investigative Dermatology, 2019, 139, 244-249.	0.7	23
44	Bazexâ€“DuprÃ©â€“Christol syndrome: An ectodermal dysplasia with skin appendage neoplasms. European Journal of Medical Genetics, 2009, 52, 250-255.	1.3	22
45	Trisomic rescue causing reduction to homozygosity for a novel ABCA12 mutation in harlequin ichthyosis. Clinical Genetics, 2009, 76, 392-397.	2.0	21
46	Cloning and analysis of cDNA for rat histone H1Â°. Nucleic Acids Research, 1993, 21, 1674-1674.	14.5	20
47	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
48	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
49	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
50	MicroRNAâ€145â€5p regulates fibrotic features of recessive dystrophic epidermolysis bullosa skin fibroblasts. British Journal of Dermatology, 2019, 181, 1017-1027.	1.5	19
51	Induction of senescence pathways in Kindler syndrome primary keratinocytes. British Journal of Dermatology, 2013, 168, 1019-1026.	1.5	18
52	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
53	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
54	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

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55	Expression of synapsin I gene in primary cultures of differentiating rat cortical neurons. Neurochemical Research, 1995, 20, 239-243.	3.3	16
56	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
57	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
58	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
59	Qualitative differences in nuclear proteins correlate with neuronal terminal differentiation. Cellular and Molecular Neurobiology, 1992, 12, 33-43.	3.3	15
60	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
61	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
62	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
63	A New SPINK5 Donor Splice Site Mutation in Siblings with Netherton Syndrome. Acta Dermato-Venereologica, 2010, 90, 95-96.	1.3	14
64	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
65	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
66	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
67	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
68	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
69	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
70	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
71	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
72	<scp>FERMT1</scp> promoter mutations in patients with Kindler syndrome. Clinical Genetics, 2015, 88, 248-254.	2.0	11

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73	Kindler syndrome with severe mucosal involvement in a large Palestinian pedigree. European Journal of Dermatology, 2015, 25, 14-19.	0.6	11
74	Childhood epidermolysis bullosa acquisita during squaric acid dibutyl ester immunotherapy for alopecia areata. British Journal of Dermatology, 2017, 176, 491-494.	1.5	11
75	The dynamic properties of neuronal chromatin are modulated by triiodothyronine. Neurochemical Research, 1992, 17, 1049-1055.	3.3	10
76	Dystrophic epidermolysis bullosa phenotypes in a large consanguineous Tunisian family. Journal of Dermatological Science, 2009, 54, 114-120.	1.9	10
77	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
78	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
79	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
80	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
81	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
82	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
83	AXIN2 germline mutations are rare in familial melanoma. Genes Chromosomes and Cancer, 2011, 50, 370-373.	2.8	8
84	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
85	Epidermolysis Bullosa Care in Italy. Dermatologic Clinics, 2010, 28, 407-409.	1.7	7
86	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
87	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
88	Ichthyosis Prematurity Syndrome due to a Novel SLC27A4 Homozygous Mutation in an Italian Patient. Acta Dermato-Venereologica, 2018, 98, 803-804.	1.3	7
89	Phenotypic Features of Epidermolysis Bullosa Simplex due to KLHL24 Mutations in 3 Italian Cases. Acta Dermato-Venereologica, 2019, 99, 238-239.	1.3	7
90	Mutation Mechanisms. Dermatologic Clinics, 2010, 28, 17-22.	1.7	6

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91	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
92	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
93	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
94	Mutational founder effect in recessive dystrophic epidermolysis bullosa families from Southern Tunisia. Archives of Dermatological Research, 2014, 306, 405-411.	1.9	5
95	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
96	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
97	Dystrophic epidermolysis bullosa pruriginosa with autoantibodies against collagen VII. European Journal of Dermatology, 2012, 22, 541-542.	0.6	4
98	A Postzygotic ATP2A2 Novel Mutation Identified by Next-generation Sequencing in Mosaic Darier Disease. Acta Dermato-Venereologica, 2018, 99, 115-116.	1.3	4
99	Palmoplantar Keratoderma and Woolly Hair Revealing Asymptomatic Arrhythmogenic Cardiomyopathy. Acta Dermato-Venereologica, 2019, 99, 831-832.	1.3	4
100	Development of bullous pemphigoid in junctional epidermolysis bullosa. Journal of the European Academy of Dermatology and Venereology, 2020, 34, e146-e148.	2.4	4
101	Branch point and donor splice-siteCOL7A1mutations in mild recessive dystrophic epidermolysis bullosa. British Journal of Dermatology, 2009, 161, 464-467.	1.5	3
102	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
103	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
104	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
105	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
106	Lipoid Proteinosis: A Previously Unrecognized Mutation and Therapeutic Response to Acitretin. Acta Dermato-Venereologica, 2017, 97, 1249-1251.	1.3	2
107	Accelerated features of senescence in cultured type 2 diabetic skin fibroblasts. European Journal of Dermatology, 2017, 27, 408-410.	0.6	2
108	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

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109	302 Molecular and mutational signatures of squamous cell carcinomas in epidermolysis bullosa. Journal of Investigative Dermatology, 2019, 139, S266.	0.7	1
110	Prominent Follicular Keratosis in Multiple Intestinal Atresia with Combined Immune Deficiency Caused by a TTC7A Homozygous Mutation. Genes, 2022, 13, 821.	2.4	1
111	Autoantibody Idiotypy and Neonatal B Cell Repertoire. Viral Immunology, 1989, 2, 263-269.	1.3	0
112	miRâ€145â€5p profibrotic activity in RDEB. British Journal of Dermatology, 2019, 181, e121.	1.5	0
113	RDEB ä¸šš„ miRâ€145â€5p ä¿ƒçº¤ç»´åŒ–æ´»æ€§. British Journal of Dermatology, 2019, 181, e133.	1.5	0