David P Kelsell

List of Publications by Year
in descending order

114 papers	11,118 citations	53794 45 h-index	30087 103 g-index
118 all docs	118 docs citations	118 times ranked	12574 citing authors

#	Article	IF	Citations
1	Identification of the breast cancer susceptibility gene BRCA2. Nature, 1995, 378, 789-792.	27.8	3,230
2	Germline mutations in FH predispose to dominantly inherited uterine fibroids, skin leiomyomata and papillary renal cell cancer. Nature Genetics, 2002, 30, 406-410.	21.4	1,426
3	Plectin deficiency results in muscular dystrophy with epidermolysis bullosa. Nature Genetics, 1996, 13, 450-457.	21.4	394
4	Mutations in ABCA12 Underlie the Severe Congenital Skin Disease Harlequin Ichthyosis. American Journal of Human Genetics, 2005, 76, 794-803.	6.2	302
5	Inflammatory Skin and Bowel Disease Linked toADAM17Deletion. New England Journal of Medicine, 2011, 365, 1502-1508.	27.0	285
6	Mutations in GJB6 cause hidrotic ectodermal dysplasia. Nature Genetics, 2000, 26, 142-144.	21.4	270
7	Genomic Landscape of Esophageal Squamous Cell Carcinoma inÂa Japanese Population. Gastroenterology, 2016, 150, 1171-1182.	1.3	265
8	N-terminal deletion in a desmosomal cadherin causes the autosomal dominant skin disease striate palmoplantar keratoderma. Human Molecular Genetics, 1999, 8, 971-976.	2.9	205
9	Clinical Features of Multiple Cutaneous and Uterine Leiomyomatosis. Archives of Dermatology, 2005, 141, 199-206.	1.4	176
10	The gene encoding R-spondin 4 (RSPO4), a secreted protein implicated in Wnt signaling, is mutated in inherited anonychia. Nature Genetics, 2006, 38, 1245-1247.	21.4	173
11	RHBDF2 Mutations Are Associated with Tylosis, a Familial Esophageal Cancer Syndrome. American Journal of Human Genetics, 2012, 90, 340-346.	6.2	162
12	Connexin Mutations in Skin Disease and Hearing Loss. American Journal of Human Genetics, 2001, 68, 559-568.	6.2	156
13	Cellâ€“cell connectivity: desmosomes and disease. Journal of Pathology, 2012, 226, 158-171.	4.5	153
14	p16INK4a and p14ARF Tumor Suppressor Genes Are Commonly Inactivated in Cutaneous Squamous Cell Carcinoma. Journal of Investigative Dermatology, 2004, 122, 1284-1292.	0.7	145
15	Genomewide Single Nucleotide Polymorphism Microarray Mapping in Basal Cell Carcinomas Unveils Uniparental Disomy as a Key Somatic Event. Cancer Research, 2005, 65, 8597-8603.	0.9	145
16	Harlequin Ichthyosis. Archives of Dermatology, 2011, 147, 681.	1.4	145
17	Multiple Epidermal Connexins are Expressed in Different Keratinocyte Subpopulations Including Connexin 31. Journal of Investigative Dermatology, 2001, 117, 958-964.	0.7	138
18	Intermediate filamentâ€“membrane attachments function synergistically with actin-dependent contacts to regulate intercellular adhesive strength. Journal of Cell Biology, 2002, 159, 1005-1017.	5.2	134

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19	Connexin mutations in deafness. Nature, 1998, 394, 630-631.	27.8	119
20	Keratitis?ichthyosis?deafness syndrome: disease expression and spectrum of connexin 26 (GJB2) mutations in 14 patients. British Journal of Dermatology, 2007, 156, 1015-1019.	1.5	119
21	Mutations in CSTA, Encoding Cystatin A, Underlie Exfoliative Ichthyosis and Reveal a Role for This Protease Inhibitor in Cell-Cell Adhesion. American Journal of Human Genetics, 2011, 89, 564-571.	6.2	89
22	ABCA12 Is the Major Harlequin Ichthyosis Gene. Journal of Investigative Dermatology, 2006, 126, 2408-2413.	0.7	88
23	DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. American Journal of Human Genetics, 2016, 99, 115-124.	6.2	85
24	Definition and treatment of arrhythmogenic cardiomyopathy: an updated expert panel report. European Journal of Heart Failure, 2019, 21, 955-964.	7.1	84
25	Allelic imbalances and microdeletions affecting thePTPRDgene in cutaneous squamous cell carcinomas detected using single nucleotide polymorphism microarray analysis. Genes Chromosomes and Cancer, 2007, 46, 661-669.	2.8	82
26	Mutations in AQP5, Encoding a Water-Channel Protein, Cause Autosomal-Dominant Diffuse Nonepidermolytic Palmoplantar Keratoderma. American Journal of Human Genetics, 2013, 93, 330-335.	6.2	82
27	Connexin mutations associated with palmoplantar keratoderma and profound deafness in a single family. European Journal of Human Genetics, 2000, 8, 141-144.	2.8	73
28	A mutation in GJB3 is associated with recessive erythrokeratodermia variabilis (EKV) and leads to defective trafficking of the connexin 31 protein. Human Molecular Genetics, 2002, 11, 1311-1316.	2.9	73
29	Identification of a Novel Mutation R42P in the Gap Junction Protein Î²-3 Associated with Autosomal Dominant Erythrokeratoderma Variabilis. Journal of Investigative Dermatology, 1999, 113, 1119-1122.	0.7	71
30	Early Death from Cardiomyopathy in a Family with Autosomal Dominant Striate Palmoplantar Keratoderma and Woolly Hair Associated with a Novel Insertion Mutation in Desmoplakin. Journal of Investigative Dermatology, 2006, 126, 1651-1654.	0.7	69
31	Defective trafficking and cell death is characteristic of skin disease-associated connexin 31 mutations. Human Molecular Genetics, 2002, 11, 2005-2014.	2.9	68
32	iRHOM2-dependent regulation of ADAM17 in cutaneous disease and epidermal barrier function. Human Molecular Genetics, 2014, 23, 4064-4076.	2.9	67
33	Cryptogenic multifocal ulcerating stenosing enteritis associated with homozygous deletion mutations in cytosolic phospholipase A2-Î±. Gut, 2014, 63, 96-104.	12.1	62
34	Connexins in epidermal homeostasis and skin disease. Biochimica Et Biophysica Acta - Biomembranes, 2012, 1818, 1952-1961.	2.6	61
35	Deletions in the cytoplasmic domain of iRhom1 and iRhom2 promote shedding of the TNF receptor by the protease ADAM17. Science Signaling, 2015, 8, ra109.	3.6	60
36	Clinical and Genetic Heterogeneity of Erythrokeratoderma Variabilis. Journal of Investigative Dermatology, 2005, 125, 920-927.	0.7	56

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37	Missense Mutations in Fumarate Hydratase in Multiple Cutaneous and Uterine Leiomyomatosis and Renal Cell Cancer. Journal of Molecular Diagnostics, 2005, 7, 437-443.	2.8	56
38	Tylosis with oesophageal cancer: Diagnosis, management and molecular mechanisms. Orphanet Journal of Rare Diseases, 2015, 10, 126.	2.7	55
39	The palmoplantar keratodermas: much more than palms and soles. Trends in Molecular Medicine, 1999, 5, 107-113.	2.6	53
40	Functional studies of human skin disease- and deafness-associated connexin 30 mutations. Biochemical and Biophysical Research Communications, 2002, 298, 651-656.	2.1	53
41	EKV mutant connexin 31 associated cell death is mediated by ER stress. Human Molecular Genetics, 2009, 18, 4734-4745.	2.9	53
42	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in aÂSpectrum of Keratinization Disorders Associated with Thrombocytopenia. Journal of Investigative Dermatology, 2017, 137, 2344-2353.	0.7	53
43	Premature Terminal Differentiation and a Reduction in Specific Proteases Associated with Loss of ABCA12 in Harlequin Ichthyosis. American Journal of Pathology, 2009, 174, 970-978.	3.8	51
44	Homozygous Mutations in the 5â€² Region of the JUP Gene Result in Cutaneous Disease but Normal Heart Development in Children. Journal of Investigative Dermatology, 2010, 130, 1543-1550.	0.7	49
45	Key functions for gap junctions in skin and hearing. Biochemical Journal, 2011, 438, 245-254.	3.7	49
46	A Missense Mutation in the MBTPS2 Gene Underlies the X-Linked Form of Olmsted Syndrome. Journal of Investigative Dermatology, 2013, 133, 571-573.	0.7	47
47	Connexin interaction patterns in keratinocytes revealed morphologically and by FRET analysis. Journal of Cell Science, 2005, 118, 1505-1514.	2.0	45
48	A Deafness-Associated Mutant Human Connexin 26 Improves the Epithelial Barrier In Vitro. Journal of Membrane Biology, 2007, 218, 29-37.	2.1	45
49	p63 Mediates an Apoptotic Response to Pharmacological and Disease-Related ER Stress in the Developing Epidermis. Developmental Cell, 2011, 21, 492-505.	7.0	45
50	Mutations and alternative splicing of theBRCA1 gene in UK breast/ovarian cancer families. , 1997, 18, 102-110.		43
51	Clouston hidrotic ectodermal dysplasia (HED): genetic homogeneity, presence of a founder effect in the French Canadian population and fine genetic mapping. European Journal of Human Genetics, 2000, 8, 372-380.	2.8	43
52	Mutations in R-Spondin 4 (RSPO4) Underlie Inherited Anonychia. Journal of Investigative Dermatology, 2008, 128, 867-870.	0.7	43
53	Lossâ€ofâ€function desmoplakin I and II mutations underlie dominant arrhythmogenic cardiomyopathy with a hair and skin phenotype. British Journal of Dermatology, 2019, 180, 1114-1122.	1.5	41
54	iASPP, a previously unidentified regulator of desmosomes, prevents arrhythmogenic right ventricular cardiomyopathy (ARVC)-induced sudden death. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E973-E981.	7.1	37

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55	Exome Sequencing and Rare Variant Analysis RevealsÂMultiple Filaggrin Mutations in BangladeshiÂFamilies with Atopic Eczema andÂAdditional Risk Genes. Journal of Investigative Dermatology, 2018, 138, 2674-2677.	0.7	37
56	Genetic linkage studies in non-epidermolytic palmoplantar keratoderma: evidence for heterogeneity. Human Molecular Genetics, 1995, 4, 1021-1025.	2.9	36
57	Recessive oligodontia linked to a homozygous loss-of-function mutation in the SMOC2 gene. Archives of Oral Biology, 2013, 58, 462-466.	1.8	36
58	Loss-of-Function Mutations in CAST Cause Peeling Skin, Leukonychia, Acral Punctate Keratoses, Cheilitis, and Knuckle Pads. American Journal of Human Genetics, 2015, 96, 440-447.	6.2	36
59	Rhomboid family member 2 regulates cytoskeletal stress-associated Keratin 16. Nature Communications, 2017, 8, 14174.	12.8	36
60	Properties of human connexin 31, which is implicated in hereditary dermatological disease and deafness. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 5213-5218.	7.1	31
61	3D model of harlequin ichthyosis reveals inflammatory therapeutic targets. Journal of Clinical Investigation, 2020, 130, 4798-4810.	8.2	31
62	Combined loss of BRCA1/BRCA2 in grade 3 breast carcinomas. Lancet, The, 1996, 347, 1554-1555.	13.7	28
63	Gene expression analysis of EpiDermâ„¢ following exposure to SLS using cDNA microarrays. Toxicology in Vitro, 2001, 15, 393-398.	2.4	28
64	Novel and recurring ABCA12 mutations associated with harlequin ichthyosis: implications for prenatal diagnosis. British Journal of Dermatology, 2008, 158, 611-613.	1.5	28
65	A novel M163L mutation in connexin 26 causing cell death and associated with autosomal dominant hearing loss. Hearing Research, 2008, 240, 87-92.	2.0	28
66	Insights into Desmosome Biology from Inherited Human Skin Disease and Cardiocutaneous Syndromes. Cell Communication and Adhesion, 2014, 21, 129-140.	1.0	27
67	Loss-of-Function Mutations in SERPINB8 Linked to Exfoliative Ichthyosis with Impaired Mechanical Stability of Intercellular Adhesions. American Journal of Human Genetics, 2016, 99, 430-436.	6.2	27
68	Hereditary 'white nails': a genetic and structural study. British Journal of Dermatology, 2004, 151, 65-72.	1.5	26
69	Association between loss of heterozygosity ofBRCA1 andBRCA2 and morphological attributes of sporadic breast cancer. International Journal of Cancer, 2000, 88, 204-208.	5.1	25
70	Targeted Sequence Capture and High-Throughput Sequencing in the Molecular Diagnosis of Ichthyosis and Other Skin Diseases. Journal of Investigative Dermatology, 2013, 133, 573-576.	0.7	25
71	Defective channels lead to an impaired skin barrier. Journal of Cell Science, 2014, 127, 4343-50.	2.0	25
72	The DSPII splice variant is critical for desmosome-mediated HaCaT keratinocyte adhesion. Journal of Cell Science, 2012, 125, 2853-61.	2.0	24

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73	Rhomboid proteins: a role in keratinocyte proliferation and cancer. Cell and Tissue Research, 2013, 351, 301-307.	2.9	23
74	p63 is a key regulator of iRHOM2 signalling in the keratinocyte stress response. Nature Communications, 2018, 9, 1021.	12.8	23
75	Cell Cycle- and Cancer-Associated Gene Networks Activated by Dsg2: Evidence of Cystatin A Deregulation and a Potential Role in Cell-Cell Adhesion. PLoS ONE, 2015, 10, e0120091.	2.5	22
76	Connexin 26 facilitates gastrointestinal bacterial infection in vitro. Cell and Tissue Research, 2013, 351, 107-116.	2.9	21
77	A novel de novo activating mutation in STAT3 identified in a patient with common variable immunodeficiency (CVID). Clinical Immunology, 2018, 187, 132-136.	3.2	19
78	Diagnosis and confirmation of epidermolytic palmoplantar keratoderma by the identification of mutations in keratin 9 using denaturing high-performance liquid chromatography. British Journal of Dermatology, 2002, 146, 952-957.	1.5	18
79	A novel ABCA12 mutation underlying a case of Harlequin ichthyosis. British Journal of Dermatology, 2006, 155, 204-206.	1.5	18
80	ADAM17/EGFR axis promotes transglutaminase-dependent skin barrier formation through phospholipase C Î³1 and protein kinase C pathways. Scientific Reports, 2016, 6, 39780.	3.3	18
81	Cellular Mechanisms of Mutant Connexins in Skin Disease and Hearing Loss. Cell Communication and Adhesion, 2003, 10, 347-351.	1.0	17
82	Identification and characterization of DSPIa, a novel isoform of human desmoplakin. Cell and Tissue Research, 2010, 341, 121-129.	2.9	17
83	Metastatic cutaneous squamous cell carcinoma shows frequent deletion in the protein tyrosine phosphatase receptor Type D gene. International Journal of Cancer, 2012, 131, E216-26.	5.1	17
84	An autosomal recessive exfoliative ichthyosis with linkage to chromosome 12q13. British Journal of Dermatology, 2003, 149, 174-180.	1.5	16
85	Connexin mutations in human disease. Experimental Dermatology, 2004, 13, 661-662.	2.9	15
86	Tissue-specific effects of wild-type and mutant connexin 31: a role in neurite outgrowth. Human Molecular Genetics, 2007, 16, 165-172.	2.9	14
87	Filaggrin mutations are associated with ichthyosis vulgaris in the Bangladeshi population. British Journal of Dermatology, 2009, 160, 1113-1115.	1.5	14
88	A novel frameshift MSX1 mutation in a Saudi family with autosomal dominant premolar and third molar agenesis. Archives of Oral Biology, 2015, 60, 982-988.	1.8	12
89	A Novel Mechanism for Activation of GLI1 by Nuclear SMO That Escapes Anti-SMO Inhibitors. Cancer Research, 2018, 78, 2577-2588.	0.9	12
90	R-Spondins in Cutaneous Biology: Nails and Cancer. Cell Cycle, 2007, 6, 895-897.	2.6	11

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91	SNPing at the Epidermal Barrier. Journal of Investigative Dermatology, 2011, 131, 1593-1595.	0.7	11
92	Discovery in Genetic Skin Disease: The Impact of High Throughput Genetic Technologies. Genes, 2014, 5, 615-634.	2.4	10
93	Mutations inEDAandEDARGenes in a Large Mexican Hispanic Cohort with Hypohidrotic Ectodermal Dysplasia. Annals of Dermatology, 2015, 27, 474.	0.9	10
94	Cardiomyopathy diagnosed in the eldest child harbouring p.S24X mutation inJUP. British Journal of Dermatology, 2016, 175, 644-646.	1.5	10
95	Recessive Mutation in FAM83G Associated with Palmoplantar Keratoderma and Exuberant Scalp Hair. Journal of Investigative Dermatology, 2018, 138, 984-987.	0.7	10
96	New ANTXR1 Gene Mutation for GAPO Syndrome: A Case Report. Molecular Syndromology, 2016, 7, 160-163.	0.8	9
97	Cellular biomechanics impairment in keratinocytes is associated with a C-terminal truncated desmoplakin: An atomic force microscopy investigation. Micron, 2018, 106, 27-33.	2.2	8
98	iRHOM2: A Regulator of Palmoplantar Biology, Inflammation, and Viral Susceptibility. Journal of Investigative Dermatology, 2021, 141, 722-726.	0.7	7
99	Exoming into Rare Skin Disease: EGFR Deficiency. Journal of Investigative Dermatology, 2014, 134, 2486-2488.	0.7	6
100	Role for WNT16B in human epidermal keratinocyte proliferation and differentiation. Journal of Cell Science, 2007, 120, 917-917.	2.0	4
101	Novel ABCA12 mutations in harlequin ichthyosis: A journey from photo diagnosis to prenatal diagnosis. Gene, 2015, 556, 254-256.	2.2	3
102	A severe collodion phenotype in the newborn period associated with a homozygous missense mutation inALOX12B. British Journal of Dermatology, 2015, 173, 285-287.	1.5	2
103	Evolution of Electrocardiographic and Structural Features Over 3 Decades in Arrhythmogenic Cardiomyopathy. Circulation, 2015, 131, 2233-2235.	1.6	2
104	A profile of lipid dysregulation in harlequin ichthyosis. British Journal of Dermatology, 2017, 177, e217-e219.	1.5	2
105	Clinical variability of the GJB4:c.35GÂ>ÂA gene variant: a study of a large Brazilian erythrokeratodermia pedigree. International Journal of Dermatology, 2020, 59, 722-725.	1.0	2
106	Connexins in Skin Biology. , 2009, , 307-321.		2
107	Double jeopardy: Ras and CDK4 co-expression in skin cancer. Trends in Molecular Medicine, 2002, 8, 548.	6.7	1
108	Cover image: Unpeeling the layers of harlequin ichthyosis. British Journal of Dermatology, 2016, 174, 1160-1161.	1.5	1

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109	Whats new in genodermatoses?. Keio Journal of Medicine, 2001, 50, 35-38.	1.1	1
110	Modelling of temporal exposure to the ambient environment and eczema severity. JID Innovations, 2021, 2, 100062.	2.4	1
111	Identifying Mutations in Single Gene Disorders. , 0, , 145-164.		0
112	Current insights into protease dynamics in human epithelial disease and barrier function. Cell and Tissue Research, 2013, 351, 213-215.	2.9	0
113	Celebrating the 50th Anniversary of ESDR. Journal of Investigative Dermatology, 2020, 140, S145-S146.	0.7	0
114	The Future of ESDR. Journal of Investigative Dermatology, 2020, 140, S192-S193.	0.7	0