

David P Kelsell

List of Publications by Year in descending order

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114
papers

11,118
citations

53660

45
h-index

30010

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. <i>Nature</i> , 1995, 378, 789-792.	13.7	3,230
2	Germline mutations in FH predispose to dominantly inherited uterine fibroids, skin leiomyomata and papillary renal cell cancer. <i>Nature Genetics</i> , 2002, 30, 406-410.	9.4	1,426
3	Plectin deficiency results in muscular dystrophy with epidermolysis bullosa. <i>Nature Genetics</i> , 1996, 13, 450-457.	9.4	394
4	Mutations in ABCA12 Underlie the Severe Congenital Skin Disease Harlequin Ichthyosis. <i>American Journal of Human Genetics</i> , 2005, 76, 794-803.	2.6	302
5	Inflammatory Skin and Bowel Disease Linked to ADAM17 Deletion. <i>New England Journal of Medicine</i> , 2011, 365, 1502-1508.	13.9	285
6	Mutations in GJB6 cause hidrotic ectodermal dysplasia. <i>Nature Genetics</i> , 2000, 26, 142-144.	9.4	270
7	Genomic Landscape of Esophageal Squamous Cell Carcinoma in a Japanese Population. <i>Gastroenterology</i> , 2016, 150, 1171-1182.	0.6	265
8	N-terminal deletion in a desmosomal cadherin causes the autosomal dominant skin disease striate palmoplantar keratoderma. <i>Human Molecular Genetics</i> , 1999, 8, 971-976.	1.4	205
9	Clinical Features of Multiple Cutaneous and Uterine Leiomyomatosis. <i>Archives of Dermatology</i> , 2005, 141, 199-206.	1.7	176
10	The gene encoding R-spondin 4 (RSPO4), a secreted protein implicated in Wnt signaling, is mutated in inherited anonychia. <i>Nature Genetics</i> , 2006, 38, 1245-1247.	9.4	173
11	RHBDF2 Mutations Are Associated with Tylosis, a Familial Esophageal Cancer Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 340-346.	2.6	162
12	Connexin Mutations in Skin Disease and Hearing Loss. <i>American Journal of Human Genetics</i> , 2001, 68, 559-568.	2.6	156
13	Cell-cell connectivity: desmosomes and disease. <i>Journal of Pathology</i> , 2012, 226, 158-171.	2.1	153
14	p16INK4a and p14ARF Tumor Suppressor Genes Are Commonly Inactivated in Cutaneous Squamous Cell Carcinoma. <i>Journal of Investigative Dermatology</i> , 2004, 122, 1284-1292.	0.3	145
15	Genomewide Single Nucleotide Polymorphism Microarray Mapping in Basal Cell Carcinomas Unveils Uniparental Disomy as a Key Somatic Event. <i>Cancer Research</i> , 2005, 65, 8597-8603.	0.4	145
16	Harlequin Ichthyosis. <i>Archives of Dermatology</i> , 2011, 147, 681.	1.7	145
17	Multiple Epidermal Connexins are Expressed in Different Keratinocyte Subpopulations Including Connexin 31. <i>Journal of Investigative Dermatology</i> , 2001, 117, 958-964.	0.3	138
18	Intermediate filament-membrane attachments function synergistically with actin-dependent contacts to regulate intercellular adhesive strength. <i>Journal of Cell Biology</i> , 2002, 159, 1005-1017.	2.3	134

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19	Connexin mutations in deafness. <i>Nature</i> , 1998, 394, 630-631.	13.7	119
20	Keratitis?ichthyosis?deafness syndrome: disease expression and spectrum of connexin 26 (GJB2) mutations in 14 patients. <i>British Journal of Dermatology</i> , 2007, 156, 1015-1019.	1.4	119
21	Mutations in CSTA, Encoding Cystatin A, Underlie Exfoliative Ichthyosis and Reveal a Role for This Protease Inhibitor in Cell-Cell Adhesion. <i>American Journal of Human Genetics</i> , 2011, 89, 564-571.	2.6	89
22	ABCA12 Is the Major Harlequin Ichthyosis Gene. <i>Journal of Investigative Dermatology</i> , 2006, 126, 2408-2413.	0.3	88
23	DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. <i>American Journal of Human Genetics</i> , 2016, 99, 115-124.	2.6	85
24	Definition and treatment of arrhythmogenic cardiomyopathy: an updated expert panel report. <i>European Journal of Heart Failure</i> , 2019, 21, 955-964.	2.9	84
25	Allelic imbalances and microdeletions affecting thePTPRDgene in cutaneous squamous cell carcinomas detected using single nucleotide polymorphism microarray analysis. <i>Genes Chromosomes and Cancer</i> , 2007, 46, 661-669.	1.5	82
26	Mutations in AQP5, Encoding a Water-Channel Protein, Cause Autosomal-Dominant Diffuse Nonepidermolytic Palmoplantar Keratoderma. <i>American Journal of Human Genetics</i> , 2013, 93, 330-335.	2.6	82
27	Connexin mutations associated with palmoplantar keratoderma and profound deafness in a single family. <i>European Journal of Human Genetics</i> , 2000, 8, 141-144.	1.4	73
28	A mutation in GJB3 is associated with recessive erythrokeratoderma variabilis (EKV) and leads to defective trafficking of the connexin 31 protein. <i>Human Molecular Genetics</i> , 2002, 11, 1311-1316.	1.4	73
29	Identification of a Novel Mutation R42P in the Gap Junction Protein γ 2-3 Associated with Autosomal Dominant Erythrokeratoderma Variabilis. <i>Journal of Investigative Dermatology</i> , 1999, 113, 1119-1122.	0.3	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. <i>Journal of Investigative Dermatology</i> , 2006, 126, 1651-1654.	0.3	69
31	Defective trafficking and cell death is characteristic of skin disease-associated connexin 31 mutations. <i>Human Molecular Genetics</i> , 2002, 11, 2005-2014.	1.4	68
32	iRHOM2-dependent regulation of ADAM17 in cutaneous disease and epidermal barrier function. <i>Human Molecular Genetics</i> , 2014, 23, 4064-4076.	1.4	67
33	Cryptogenic multifocal ulcerating stenosing enteritis associated with homozygous deletion mutations in cytosolic phospholipase A2- β . <i>Gut</i> , 2014, 63, 96-104.	6.1	62
34	Connexins in epidermal homeostasis and skin disease. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 2012, 1818, 1952-1961.	1.4	61
35	Deletions in the cytoplasmic domain of iRhom1 and iRhom2 promote shedding of the TNF receptor by the protease ADAM17. <i>Science Signaling</i> , 2015, 8, ra109.	1.6	60
36	Clinical and Genetic Heterogeneity of Erythrokeratoderma Variabilis. <i>Journal of Investigative Dermatology</i> , 2005, 125, 920-927.	0.3	56

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37	Missense Mutations in Fumarate Hydratase in Multiple Cutaneous and Uterine Leiomyomatosis and Renal Cell Cancer. <i>Journal of Molecular Diagnostics</i> , 2005, 7, 437-443.	1.2	56
38	Tylosis with oesophageal cancer: Diagnosis, management and molecular mechanisms. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 126.	1.2	55
39	The palmoplantar keratodermas: much more than palms and soles. <i>Trends in Molecular Medicine</i> , 1999, 5, 107-113.	2.6	53
40	Functional studies of human skin disease- and deafness-associated connexin 30 mutations. <i>Biochemical and Biophysical Research Communications</i> , 2002, 298, 651-656.	1.0	53
41	EKV mutant connexin 31 associated cell death is mediated by ER stress. <i>Human Molecular Genetics</i> , 2009, 18, 4734-4745.	1.4	53
42	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in a Spectrum of Keratinization Disorders Associated with Thrombocytopenia. <i>Journal of Investigative Dermatology</i> , 2017, 137, 2344-2353.	0.3	53
43	Premature Terminal Differentiation and a Reduction in Specific Proteases Associated with Loss of ABCA12 in Harlequin Ichthyosis. <i>American Journal of Pathology</i> , 2009, 174, 970-978.	1.9	51
44	Homozygous Mutations in the 5' Region of the JUP Gene Result in Cutaneous Disease but Normal Heart Development in Children. <i>Journal of Investigative Dermatology</i> , 2010, 130, 1543-1550.	0.3	49
45	Key functions for gap junctions in skin and hearing. <i>Biochemical Journal</i> , 2011, 438, 245-254.	1.7	49
46	A Missense Mutation in the MBTPS2 Gene Underlies the X-Linked Form of Olmsted Syndrome. <i>Journal of Investigative Dermatology</i> , 2013, 133, 571-573.	0.3	47
47	Connexin interaction patterns in keratinocytes revealed morphologically and by FRET analysis. <i>Journal of Cell Science</i> , 2005, 118, 1505-1514.	1.2	45
48	A Deafness-Associated Mutant Human Connexin 26 Improves the Epithelial Barrier In Vitro. <i>Journal of Membrane Biology</i> , 2007, 218, 29-37.	1.0	45
49	p63 Mediates an Apoptotic Response to Pharmacological and Disease-Related ER Stress in the Developing Epidermis. <i>Developmental Cell</i> , 2011, 21, 492-505.	3.1	45
50	Mutations and alternative splicing of the BRCA1 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. <i>European Journal of Human Genetics</i> , 2000, 8, 372-380.	1.4	43
52	Mutations in R-Spondin 4 (RSPO4) Underlie Inherited Anonychia. <i>Journal of Investigative Dermatology</i> , 2008, 128, 867-870.	0.3	43
53	Loss of function desmoplakin I and II mutations underlie dominant arrhythmogenic cardiomyopathy with a hair and skin phenotype. <i>British Journal of Dermatology</i> , 2019, 180, 1114-1122.	1.4	41
54	iASPP, a previously unidentified regulator of desmosomes, prevents arrhythmogenic right ventricular cardiomyopathy (ARVC)-induced sudden death. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E973-E981.	3.3	37

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55	Exome Sequencing and Rare Variant Analysis Reveals Multiple Filaggrin Mutations in Bangladeshi Families with Atopic Eczema and Additional Risk Genes. <i>Journal of Investigative Dermatology</i> , 2018, 138, 2674-2677.	0.3	37
56	Genetic linkage studies in non-epidermolytic palmoplantar keratoderma: evidence for heterogeneity. <i>Human Molecular Genetics</i> , 1995, 4, 1021-1025.	1.4	36
57	Recessive oligodontia linked to a homozygous loss-of-function mutation in the <i>SMOC2</i> gene. <i>Archives of Oral Biology</i> , 2013, 58, 462-466.	0.8	36
58	Loss-of-Function Mutations in <i>CAST</i> Cause Peeling Skin, Leukonychia, Acral Punctate Keratoses, Cheilitis, and Knuckle Pads. <i>American Journal of Human Genetics</i> , 2015, 96, 440-447.	2.6	36
59	Rhomboid family member 2 regulates cytoskeletal stress-associated Keratin 16. <i>Nature Communications</i> , 2017, 8, 14174.	5.8	36
60	Properties of human connexin 31, which is implicated in hereditary dermatological disease and deafness. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 5213-5218.	3.3	31
61	3D model of harlequin ichthyosis reveals inflammatory therapeutic targets. <i>Journal of Clinical Investigation</i> , 2020, 130, 4798-4810.	3.9	31
62	Combined loss of <i>BRCA1/BRCA2</i> in grade 3 breast carcinomas. <i>Lancet</i> , The, 1996, 347, 1554-1555.	6.3	28
63	Gene expression analysis of EpiDerm [®] following exposure to SLS using cDNA microarrays. <i>Toxicology in Vitro</i> , 2001, 15, 393-398.	1.1	28
64	Novel and recurring <i>ABCA12</i> mutations associated with harlequin ichthyosis: implications for prenatal diagnosis. <i>British Journal of Dermatology</i> , 2008, 158, 611-613.	1.4	28
65	A novel M163L mutation in connexin 26 causing cell death and associated with autosomal dominant hearing loss. <i>Hearing Research</i> , 2008, 240, 87-92.	0.9	28
66	Insights into Desmosome Biology from Inherited Human Skin Disease and Cardiocutaneous Syndromes. <i>Cell Communication and Adhesion</i> , 2014, 21, 129-140.	1.0	27
67	Loss-of-Function Mutations in <i>SERPIN8</i> Linked to Exfoliative Ichthyosis with Impaired Mechanical Stability of Intercellular Adhesions. <i>American Journal of Human Genetics</i> , 2016, 99, 430-436.	2.6	27
68	Hereditary 'white nails': a genetic and structural study. <i>British Journal of Dermatology</i> , 2004, 151, 65-72.	1.4	26
69	Association between loss of heterozygosity of <i>BRCA1</i> and <i>BRCA2</i> and morphological attributes of sporadic breast cancer. <i>International Journal of Cancer</i> , 2000, 88, 204-208.	2.3	25
70	Targeted Sequence Capture and High-Throughput Sequencing in the Molecular Diagnosis of Ichthyosis and Other Skin Diseases. <i>Journal of Investigative Dermatology</i> , 2013, 133, 573-576.	0.3	25
71	Defective channels lead to an impaired skin barrier. <i>Journal of Cell Science</i> , 2014, 127, 4343-50.	1.2	25
72	The <i>DSP11</i> splice variant is critical for desmosome-mediated HaCaT keratinocyte adhesion. <i>Journal of Cell Science</i> , 2012, 125, 2853-61.	1.2	24

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73	Rhomboid proteins: a role in keratinocyte proliferation and cancer. <i>Cell and Tissue Research</i> , 2013, 351, 301-307.	1.5	23
74	p63 is a key regulator of iRHOM2 signalling in the keratinocyte stress response. <i>Nature Communications</i> , 2018, 9, 1021.	5.8	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. <i>PLoS ONE</i> , 2015, 10, e0120091.	1.1	22
76	Connexin 26 facilitates gastrointestinal bacterial infection in vitro. <i>Cell and Tissue Research</i> , 2013, 351, 107-116.	1.5	21
77	A novel de novo activating mutation in STAT3 identified in a patient with common variable immunodeficiency (CVID). <i>Clinical Immunology</i> , 2018, 187, 132-136.	1.4	19
78	Diagnosis and confirmation of epidermolytic palmoplantar keratoderma by the identification of mutations in keratin 9 using denaturing high-performance liquid chromatography. <i>British Journal of Dermatology</i> , 2002, 146, 952-957.	1.4	18
79	A novel ABCA12 mutation underlying a case of Harlequin ichthyosis. <i>British Journal of Dermatology</i> , 2006, 155, 204-206.	1.4	18
80	ADAM17/EGFR axis promotes transglutaminase-dependent skin barrier formation through phospholipase C β 1 and protein kinase C pathways. <i>Scientific Reports</i> , 2016, 6, 39780.	1.6	18
81	Cellular Mechanisms of Mutant Connexins in Skin Disease and Hearing Loss. <i>Cell Communication and Adhesion</i> , 2003, 10, 347-351.	1.0	17
82	Identification and characterization of DSPIa, a novel isoform of human desmoplakin. <i>Cell and Tissue Research</i> , 2010, 341, 121-129.	1.5	17
83	Metastatic cutaneous squamous cell carcinoma shows frequent deletion in the protein tyrosine phosphatase receptor Type D gene. <i>International Journal of Cancer</i> , 2012, 131, E216-26.	2.3	17
84	An autosomal recessive exfoliative ichthyosis with linkage to chromosome 12q13. <i>British Journal of Dermatology</i> , 2003, 149, 174-180.	1.4	16
85	Connexin mutations in human disease. <i>Experimental Dermatology</i> , 2004, 13, 661-662.	1.4	15
86	Tissue-specific effects of wild-type and mutant connexin 31: a role in neurite outgrowth. <i>Human Molecular Genetics</i> , 2007, 16, 165-172.	1.4	14
87	Filaggrin mutations are associated with ichthyosis vulgaris in the Bangladeshi population. <i>British Journal of Dermatology</i> , 2009, 160, 1113-1115.	1.4	14
88	A novel frameshift MSX1 mutation in a Saudi family with autosomal dominant premolar and third molar agenesis. <i>Archives of Oral Biology</i> , 2015, 60, 982-988.	0.8	12
89	A Novel Mechanism for Activation of GLI1 by Nuclear SMO That Escapes Anti-SMO Inhibitors. <i>Cancer Research</i> , 2018, 78, 2577-2588.	0.4	12
90	R-Spondins in Cutaneous Biology: Nails and Cancer. <i>Cell Cycle</i> , 2007, 6, 895-897.	1.3	11

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91	SNPing at the Epidermal Barrier. <i>Journal of Investigative Dermatology</i> , 2011, 131, 1593-1595.	0.3	11
92	Discovery in Genetic Skin Disease: The Impact of High Throughput Genetic Technologies. <i>Genes</i> , 2014, 5, 615-634.	1.0	10
93	Mutations in EDA and EDAR Genes in a Large Mexican Hispanic Cohort with Hypohidrotic Ectodermal Dysplasia. <i>Annals of Dermatology</i> , 2015, 27, 474.	0.3	10
94	Cardiomyopathy diagnosed in the eldest child harbouring p.S24X mutation in <i>JUP</i> . <i>British Journal of Dermatology</i> , 2016, 175, 644-646.	1.4	10
95	Recessive Mutation in FAM83G Associated with Palmoplantar Keratoderma and Exuberant Scalp Hair. <i>Journal of Investigative Dermatology</i> , 2018, 138, 984-987.	0.3	10
96	New ANTXR1 Gene Mutation for GAPO Syndrome: A Case Report. <i>Molecular Syndromology</i> , 2016, 7, 160-163.	0.3	9
97	Cellular biomechanics impairment in keratinocytes is associated with a C-terminal truncated desmoplakin: An atomic force microscopy investigation. <i>Micron</i> , 2018, 106, 27-33.	1.1	8
98	iRHOM2: A Regulator of Palmoplantar Biology, Inflammation, and Viral Susceptibility. <i>Journal of Investigative Dermatology</i> , 2021, 141, 722-726.	0.3	7
99	Exoming into Rare Skin Disease: EGFR Deficiency. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2486-2488.	0.3	6
100	Role for WNT16B in human epidermal keratinocyte proliferation and differentiation. <i>Journal of Cell Science</i> , 2007, 120, 917-917.	1.2	4
101	Novel ABCA12 mutations in harlequin ichthyosis: A journey from photo diagnosis to prenatal diagnosis. <i>Gene</i> , 2015, 556, 254-256.	1.0	3
102	A severe collodion phenotype in the newborn period associated with a homozygous missense mutation in <i>ALOX12B</i> . <i>British Journal of Dermatology</i> , 2015, 173, 285-287.	1.4	2
103	Evolution of Electrocardiographic and Structural Features Over 3 Decades in Arrhythmogenic Cardiomyopathy. <i>Circulation</i> , 2015, 131, 2233-2235.	1.6	2
104	A profile of lipid dysregulation in harlequin ichthyosis. <i>British Journal of Dermatology</i> , 2017, 177, e217-e219.	1.4	2
105	Clinical variability of the <i>GJB4:c.35G>A</i> gene variant: a study of a large Brazilian erythrokeratoderma pedigree. <i>International Journal of Dermatology</i> , 2020, 59, 722-725.	0.5	2
106	Connexins in Skin Biology. , 2009, , 307-321.		2
107	Double jeopardy: Ras and CDK4 co-expression in skin cancer. <i>Trends in Molecular Medicine</i> , 2002, 8, 548.	3.5	1
108	Cover image: Unpeeling the layers of harlequin ichthyosis. <i>British Journal of Dermatology</i> , 2016, 174, 1160-1161.	1.4	1

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109	Whats new in genodermatoses?. Keio Journal of Medicine, 2001, 50, 35-38.	0.5	1
110	Modelling of temporal exposure to the ambient environment and eczema severity. JID Innovations, 2021, 2, 100062.	1.2	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.	1.5	0
113	Celebrating the 50th Anniversary of ESDR. Journal of Investigative Dermatology, 2020, 140, S145-S146.	0.3	0
114	The Future of ESDR. Journal of Investigative Dermatology, 2020, 140, S192-S193.	0.3	0