John S Mcgrath

List of Publications by Year
in descending order

562 papers	26,443 citations	6760 80 h-index	13250 132 g-index
604 all docs	604 docs citations	604 times ranked	32258 citing authors

#	Article	IF	Citations
1	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, 931-950.	1.2	823
2	Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification. Journal of the American Academy of Dermatology, 2014, 70, 1103-1126.	1.2	762
3	Mutations in the Tight-Junction Gene Claudin 19 (CLDN19) Are Associated with Renal Magnesium Wasting, Renal Failure, and Severe Ocular Involvement. American Journal of Human Genetics, 2006, 79, 949-957.	6.1	454
4	Alopecia Universalis Associated with a Mutation in the Human hairless Gene. Science, 1998, 279, 720-724.	20.9	433
5	Prediction of Real-World Functional Disability in Chronic Mental Disorders: A Comparison of Schizophrenia and Bipolar Disorder. American Journal of Psychiatry, 2010, 167, 1116-1124.	8.7	403
6	Revised classification system for inherited epidermolysis bullosa. Journal of the American Academy of Dermatology, 2000, 42, 1051-1066.	1.2	394
7	Mutations in the 180â€“kD bullous pemphigoid antigen (BPAG2), a hemidesmosomal transmembrane collagen (COL17A1), in generalized atrophic benign epidermolysis bullosa. Nature Genetics, 1995, 11, 83-86.	20.4	370
8	Mutations in the plakophilin 1 gene result in ectodermal dysplasia/skin fragility syndrome. Nature Genetics, 1997, 17, 240-244.	20.4	366
9	Bipolar I Disorder and Schizophrenia: A 440â€“Single-Nucleotide Polymorphism Screen of 64 Candidate Genes among Ashkenazi Jewish Case-Parent Trios. American Journal of Human Genetics, 2005, 77, 918-936.	6.1	360
10	Bone Marrow Transplantation for Recessive Dystrophic Epidermolysis Bullosa. New England Journal of Medicine, 2010, 363, 629-639.	30.1	332
11	Germline NLRP1 Mutations Cause Skin Inflammatory and Cancer Susceptibility Syndromes via Inflammasome Activation. Cell, 2016, 167, 187-202.e17.	27.8	331
12	Loss of Kindlin-1, a Human Homolog of the Caenorhabditis elegans Actinâ€“Extracellular-Matrix Linker Protein UNC-112, Causes Kindler Syndrome. American Journal of Human Genetics, 2003, 73, 174-187.	6.1	308
13	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. Nature Genetics, 2013, 45, 1244-1248.	20.4	298
14	Autoantibodies to extracellular matrix protein 1 in lichen sclerosus. Lancet, The, 2003, 362, 118-123.	12.1	293
15	Microstructure of cryogenic treated M2 tool steel. Materials Science & Engineering A: Structural Materials: Properties, Microstructure and Processing, 2003, 339, 241-244.	5.6	252
16	Multicenter Assessment of Neoadjuvant Chemotherapy for Muscle-invasive Bladder Cancer. European Urology, 2015, 67, 241-249.	5.0	246
17	A molecular defect in loricrin, the major component of the cornified cell envelope, underlies Vohwinkel's syndrome. Nature Genetics, 1996, 13, 70-77.	20.4	237
18	PDGFRÎ±-positive cells in bone marrow are mobilized by high mobility group box 1 (HMGB1) to regenerate injured epithelia. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 6609-6614.	7.6	223

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19	Potential of Fibroblast Cell Therapy for Recessive Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2008, 128, 2179-2189.	0.7	222
20	Microdeletions of 3q29 Confer High Risk for Schizophrenia. American Journal of Human Genetics, 2010, 87, 229-236.	6.1	219
21	Mutations in the Gene Encoding Capillary Morphogenesis Protein 2 Cause Juvenile Hyaline Fibromatosis and Infantile Systemic Hyalinosis. American Journal of Human Genetics, 2003, 73, 791-800.	6.1	215
22	Genomeâ€wide association study of schizophrenia in Ashkenazi Jews. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2015, 168, 649-659.	1.9	209
23	Interleukin-11 is a therapeutic target in idiopathic pulmonary fibrosis. Science Translational Medicine, 2019, 11, .	13.4	208
24	Epidermolysis Bullosa Simplex (Dowling-Meara Type) Is a Genetic Disease Characterized by an Abnormal Keratin-Filament Network Involving Keratins K5 and K14. Journal of Investigative Dermatology, 1991, 97, 959-968.	0.7	203
25	The filaggrin story: novel insights into skin-barrier function and disease. Trends in Molecular Medicine, 2008, 14, 20-27.	7.1	199
26	Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: Results from the Initial Screening Round of the IMPACT Study. European Urology, 2014, 66, 489-499.	5.0	198
27	Introduction of an enhanced recovery protocol for radical cystectomy. BJU International, 2008, 101, 698-701.	2.8	180
28	Resistance to 2',3'-dideoxycytidine conferred by a mutation in codon 65 of the human immunodeficiency virus type 1 reverse transcriptase. Antimicrobial Agents and Chemotherapy, 1994, 38, 282-287.	3.4	172
29	Phenotype, Genotype, and Sustained Response to Anakinra in 22 Patients With Autoinflammatory Disease Associated With CIAS-1/NALP3 Mutations. Archives of Dermatology, 2006, 142, 1591-7.	1.4	171
30	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842.	5.0	161
31	The Molecular Pathology of Progressive Symmetric Erythrokeratoderma: A Frameshift Mutation in the Loricrin Gene and Perturbations in the Cornified Cell Envelope. American Journal of Human Genetics, 1997, 61, 581-589.	6.1	157
32	The molecular basis of lipoid proteinosis: mutations in extracellular matrix protein 1. Experimental Dermatology, 2007, 16, 881-890.	2.9	152
33	Multilevel resistive switching in Ti/CuxO/Pt memory devices. Journal of Applied Physics, 2010, 108, .	2.3	147
34	LEKTI Is Localized in Lamellar Granules, Separated from KLK5 and KLK7, and Is Secreted in the Extracellular Spaces of the Superficial Stratum Granulosum. Journal of Investigative Dermatology, 2005, 124, 360-366.	0.7	142
35	Effect of Robot-Assisted Radical Cystectomy With Intracorporeal Urinary Diversion vs Open Radical Cystectomy on 90-Day Morbidity and Mortality Among Patients With Bladder Cancer. JAMA - Journal of the American Medical Association, 2022, 327, 2092.	7.0	141
36	A Homozygous Nonsense Mutation within the Dystonin Gene Coding for the Coiled-Coil Domain of the Epithelial Isoform of BPAG1 Underlies a New Subtype of Autosomal Recessive Epidermolysis Bullosa Simplex. Journal of Investigative Dermatology, 2010, 130, 1551-1557.	0.7	136

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37	Genetic Diseases of Junctions. Journal of Investigative Dermatology, 2007, 127, 2713-2725.	0.7	133
38	Altered Laminin 5 Expression Due to Mutations in the Gene Encoding the Î²3 Chain (LAMB3) in Generalized Atrophic Benign Epidermolysis Bullosa. Journal of Investigative Dermatology, 1995, 104, 467-474.	0.7	132
39	Striate Palmoplantar Keratoderma Resulting from Desmoplakin Haploinsufficiency. Journal of Investigative Dermatology, 1999, 113, 940-946.	0.7	131
40	Selective Involvement of Keratins K1 and K10 in the Cytoskeletal Abnormality of Epidermolytic Hyperkeratosis (Bullous Congenital Ichthyosiform Erythroderma). Journal of Investigative Dermatology, 1992, 99, 19-26.	0.7	129
41	Compound Heterozygosity for Non-Sense and Mis-Sense Mutations in Desmoplakin Underlies Skin Fragility/Woolly Hair Syndrome. Journal of Investigative Dermatology, 2002, 118, 232-238.	0.7	129
42	Lichen planus and lichenoid dermatoses. Journal of the American Academy of Dermatology, 2018, 79, 789-804.	1.2	127
43	Structural Variations in Anchoring Fibrils in Dystrophic Epidermolysis Bullosa: Correlation with Type VII Collagen Expression. Journal of Investigative Dermatology, 1993, 100, 366-372.	0.7	125
44	An unusual N-terminal deletion of the laminin Â3a isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. Human Molecular Genetics, 2003, 12, 2395-2409.	3.0	124
45	Extracellular Matrix Protein 1 Gene (ECM1) Mutations in Lipoid Proteinosis and Genotype-Phenotype Correlation. Journal of Investigative Dermatology, 2003, 120, 345-350.	0.7	123
46	Oncostatin M Receptor-Î² Mutations Underlie Familial Primary Localized Cutaneous Amyloidosis. American Journal of Human Genetics, 2008, 82, 73-80.	6.1	123
47	Enhanced Recovery After Robot-assisted Radical Cystectomy: EAU Robotic Urology Section Scientific Working Group Consensus View. European Urology, 2016, 70, 649-660.	5.0	121
48	Naegeli-Franceschetti-Jadassohn Syndrome and Dermatopathia Pigmentosa Reticularis: Two Allelic Ectodermal Dysplasias Caused by Dominant Mutations in KRT14. American Journal of Human Genetics, 2006, 79, 724-730.	6.1	116
49	Kindlin-1 controls Wnt and TGF-Î² availability to regulate cutaneous stem cell proliferation. Nature Medicine, 2014, 20, 350-359.	30.1	114
50	WNT10A mutation causes ectodermal dysplasia by impairing progenitor cell proliferation and KLF4-mediated differentiation. Nature Communications, 2017, 8, 15397.	13.2	111
51	Fibroblast-Derived Dermal Matrix Drives Development of Aggressive Cutaneous Squamous Cell Carcinoma in Patients with Recessive Dystrophic Epidermolysis Bullosa. Cancer Research, 2012, 72, 3522-3534.	0.9	110
52	Relationship of the Brief UCSD Performanceâ€based Skills Assessment (UPSAâ€B) to multiple indicators of functioning in people with schizophrenia and bipolar disorder. Bipolar Disorders, 2010, 12, 45-55.	2.5	108
53	Revertant mosaicism in skin: natural gene therapy. Trends in Molecular Medicine, 2011, 17, 140-148.	7.1	106
54	GATA6 Plays an Important Role in the Induction of Human Definitive Endoderm, Development of the Pancreas, and Functionality of Pancreatic Î²ÂCells. Stem Cell Reports, 2017, 8, 589-604.	4.7	106

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55	Autoinflammatory keratinization diseases. Journal of Allergy and Clinical Immunology, 2017, 140, 1545-1547.	2.9	105
56	Mutational Hotspots in the LAMB3 Gene in the Lethal (Herlitz) Type of Junctional Epidermolysis Bullosa. Human Molecular Genetics, 1996, 5, 231-237.	3.0	103
57	3D InÂVitro Model of a Functional Epidermal Permeability Barrier from Human Embryonic Stem Cells and Induced Pluripotent Stem Cells. Stem Cell Reports, 2014, 2, 675-689.	4.7	102
58	Radiofrequency-induced Thermo-chemotherapy Effect Versus a Second Course of Bacillus Calmette-GuÃ©rin or Institutional Standard in Patients with Recurrence of Nonâ€“muscle-invasive Bladder Cancer Following Induction or Maintenance Bacillus Calmette-GuÃ©rin Therapy (HYMN): A Phase III, Open-label, Randomised Controlled Trial. European Urology, 2019, 75, 63-71.	5.0	102
59	Genomewide Linkage Scan for Schizophrenia Susceptibility Loci among Ashkenazi Jewish Families Shows Evidence of Linkage on Chromosome 10q22. American Journal of Human Genetics, 2003, 73, 601-611.	6.1	101
60	LAP-VEGaS Practice Guidelines for Reporting of Educational Videos in Laparoscopic Surgery. Annals of Surgery, 2018, 268, 920-926.	4.5	100
61	APOBEC mutation drives early-onset squamous cell carcinomas in recessive dystrophic epidermolysis bullosa. Science Translational Medicine, 2018, 10, .	13.4	98
62	Increased invasive behaviour in cutaneous squamous cell carcinoma with loss of basement-membrane type VII collagen. Journal of Cell Science, 2009, 122, 1788-1799.	2.1	97
63	Frameshift Mutation in the V2 Domain of Human Keratin 1 Results in Striate Palmoplantar Keratoderma. Journal of Investigative Dermatology, 2002, 118, 838-844.	0.7	96
64	Lack of plakophilin 1 increases keratinocyte migration and reduces desmosome stability. Journal of Cell Science, 2003, 116, 3303-3314.	2.1	96
65	Activating CARD14 Mutations Are Associated with Generalized Pustular Psoriasis but Rarely Account for Familial Recurrence in Psoriasis Vulgaris. Journal of Investigative Dermatology, 2015, 135, 2964-2970.	0.7	96
66	An essential role for the Zn2+ transporter ZIP7 in B cell development. Nature Immunology, 2019, 20, 350-361.	13.9	96
67	Diseases of epidermal keratins and their linker proteins. Experimental Cell Research, 2007, 313, 1995-2009.	2.6	95
68	Limbal Stem Cell Deficiency and Ocular Phenotype in Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome Caused by p63 Mutations. Ophthalmology, 2012, 119, 74-83.	5.8	95
69	Infection and Inflammation in Schizophrenia and Bipolar Disorder: A Genome Wide Study for Interactions with Genetic Variation. PLoS ONE, 2015, 10, e0116696.	2.5	95
70	Perioperative fluid management: Consensus statement from the enhanced recovery partnership. Perioperative Medicine (London, England), 2012, 1, 2.	1.6	94
71	Keratin 9 Is Required for the Structural Integrity and Terminal Differentiation of the Palmoplantar Epidermis. Journal of Investigative Dermatology, 2014, 134, 754-763.	0.7	91
72	Novel and emerging therapies in the treatment of recessive dystrophic epidermolysis bullosa. Intractable and Rare Diseases Research, 2017, 6, 6-20.	0.9	91

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73	Extracellular matrix protein 1 inhibits the activity of matrix metalloproteinase 9 through high-affinity protein/protein interactions. Experimental Dermatology, 2006, 15, 300-307.	2.9	90
74	Risk Factors for Improper Vaccine Storage and Handling in Private Provider Offices. Pediatrics, 2001, 107, e100-e100.	2.2	88
75	Genomewide Linkage Scan for Bipolar-Disorder Susceptibility Loci among Ashkenazi Jewish Families. American Journal of Human Genetics, 2004, 75, 204-219.	6.1	88
76	Impacts of aerosols and clouds on photolysis frequencies and photochemistry during TRACE-P: 2. Three-dimensional study using a regional chemical transport model. Journal of Geophysical Research, 2003, 108, .	3.3	85
77	NFIL3/E4BP4 controls type 2 T helper cell cytokine expression. EMBO Journal, 2011, 30, 2071-2082.	8.2	84
78	Allelic Heterogeneity of Dominant and Recessive COL7A1 Mutations Underlying Epidermolysis Bullosa Pruriginosa. Journal of Investigative Dermatology, 1999, 112, 984-987.	0.7	82
79	Progress in Epidermolysis Bullosa Research: Toward Treatment and Cure. Journal of Investigative Dermatology, 2010, 130, 1778-1784.	0.7	82
80	Revised classification system for inherited epidermolysis bullosa: Report of the Second International Consensus Meeting on diagnosis and classification of epidermolysis bullosa. Journal of the American Academy of Dermatology, 2000, 42, 1051-1066.	1.2	82
81	Fine Mapping on Chromosome 10q22-q23 Implicates Neuregulin 3 in Schizophrenia. American Journal of Human Genetics, 2009, 84, 21-34.	6.1	81
82	Reporting Radical Cystectomy Outcomes Following Implementation of Enhanced Recovery After Surgery Protocols: A Systematic Review and Individual Patient Data Meta-analysis. European Urology, 2020, 78, 719-730.	5.0	81
83	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. Nature Genetics, 2012, 44, 1272-1276.	20.4	79
84	Recurrent Mutations in Kindlin-1, a Novel Keratinocyte Focal Contact Protein, in the Autosomal Recessive Skin Fragility and Photosensitivity Disorder, Kindler Syndrome. Journal of Investigative Dermatology, 2004, 122, 78-83.	0.7	78
85	Structure and function of skin, hair and nails. Medicine, 2013, 41, 317-320.	0.5	78
86	Molecular Basis of Recessive Dystrophic Epidermolysis Bullosa: Genotype/Phenotype Correlation in a Case of Moderate Clinical Severity. Journal of Investigative Dermatology, 1996, 106, 119-124.	0.7	77
87	A Missense Mutation in the Zinc-Finger Domain of the Human Hairless Gene Underlies Congenital Atrichia in a Family of Irish Travellers. American Journal of Human Genetics, 1998, 63, 984-991.	6.1	77
88	Caveolin-1 Controls Hyperresponsiveness to Mechanical Stimuli and Fibrogenesis-Associated RUNX2 ActivationÂin Keloid Fibroblasts. Journal of Investigative Dermatology, 2018, 138, 208-218.	0.7	77
89	Properties of FeSiAl-based soft magnetic composites with AlN/Al2O3 and hybrid phosphateâ€“silane insulation coatings. Journal of Alloys and Compounds, 2018, 735, 1603-1610.	5.7	77
90	Individual and Joint Effects of Early-Life Ambient PM2.5 Exposure and Maternal Prepregnancy Obesity on Childhood Overweight or Obesity. Environmental Health Perspectives, 2017, 125, 067005.	8.2	76

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91	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.	2.8	75
92	Sensitivity and specificity of the UCSD Performance-based Skills Assessment (UPSA-B) for identifying functional milestones in schizophrenia. Schizophrenia Research, 2011, 132, 165-170.	2.1	74
93	Association of obesity and treated hypertension and diabetes with cognitive ability in bipolar disorder and schizophrenia. Bipolar Disorders, 2014, 16, 422-431.	2.5	74
94	HPV-based cervical cancer screening in a population at high risk for HIV infection. International Journal of Cancer, 2000, 85, 206-210.	5.4	72
95	Familiality of Novel Factorial Dimensions of Schizophrenia. Archives of General Psychiatry, 2009, 66, 591.	13.2	72
96	Kindler Syndrome. Dermatologic Clinics, 2010, 28, 119-124.	1.8	72
97	Amlexanox Enhances Premature Termination Codon Read-Through in COL7A1 and Expression of Full Length Type VII Collagen: Potential Therapy for Recessive Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2017, 137, 1842-1849.	0.7	72
98	Robot-assisted radical cystectomy with intracorporeal urinary diversion versus open radical cystectomy (iROC): protocol for a randomised controlled trial with internal feasibility study. BMJ Open, 2018, 8, e020500.	2.1	72
99	Genomic Amplification of the Human Plakophilin 1 Gene and Detection of a New Mutation in Ectodermal Dysplasia/Skin Fragility Syndrome. Journal of Investigative Dermatology, 2000, 115, 368-374.	0.7	71
100	Patient-Specific Naturally Gene-Reverted Induced Pluripotent Stem Cells in Recessive Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2014, 134, 1246-1254.	0.7	71
101	Risk Factors in Schizophrenia. British Journal of Psychiatry, 1992, 160, 65-71.	3.6	68
102	Moderation of Phenotypic Severity in Dystrophic and Junctional Forms of Epidermolysis Bullosa Through In-Frame Skipping of Exons Containing Non-Sense or Frameshift Mutations. Journal of Investigative Dermatology, 1999, 113, 314-321.	0.7	68
103	Corneodesmosin Expression in Psoriasis Vulgaris Differs from Normal Skin and Other Inflammatory Skin Disorders. Laboratory Investigation, 2001, 81, 969-976.	3.9	68
104	Germline Mutation in ATR in Autosomal- Dominant Oropharyngeal Cancer Syndrome. American Journal of Human Genetics, 2012, 90, 511-517.	6.1	68
105	Trends in Operative Caseload and Mortality Rates after Radical Cystectomy for Bladder Cancer in England for 1998â€“2010. European Urology, 2015, 67, 1056-1062.	5.0	68
106	Frontal fibrosing alopecia: reflections and hypotheses on aetiology and pathogenesis. Experimental Dermatology, 2016, 25, 847-852.	2.9	67
107	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. American Journal of Human Genetics, 2012, 91, 1115-1121.	6.1	66
108	Pityriasis Rubra Pilaris Type V as an Autoinflammatory Disease by CARD14 Mutations. JAMA Dermatology, 2017, 153, 66.	4.2	66

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109	Autoinflammatory keratinization diseases: An emerging concept encompassing various inflammatory keratinization disorders of the skin. Journal of Dermatological Science, 2018, 90, 105-111.	2.2	66
110	Ribbon of DNA Lattice on Gold Nanoparticles for Selective Drug Delivery to Cancer Cells. Angewandte Chemie - International Edition, 2020, 59, 14584-14592.	14.8	66
111	Schizophrenia: age at onset, gender and familial risk. Acta Psychiatrica Scandinavica, 1990, 82, 344-351.	4.5	64
112	A Homozygous Deletion Mutation in the Gene Encoding the 180-kDa Bullous Pemphigoid Antigen (BPAG2) in a Family with Generalized Atrophic Benign Epidermolysis Bullosa. Journal of Investigative Dermatology, 1996, 106, 771-774.	0.7	64
113	Ectodermal Dysplasia-Skin Fragility Syndrome. Dermatologic Clinics, 2010, 28, 125-129.	1.8	64
114	Role of FAM134 paralogues in endoplasmic reticulum remodeling, ERâ€phagy, and Collagen quality control. EMBO Reports, 2021, 22, e52289.	5.1	64
115	Transplanted Bone Marrowâ€“Derived Circulating PDGFRÎ±+ Cells Restore Type VII Collagen in Recessive Dystrophic Epidermolysis Bullosa Mouse Skin Graft. Journal of Immunology, 2015, 194, 1996-2003.	0.8	63
116	Progress toward Treatment and Cure of Epidermolysis Bullosa: Summary of the DEBRA International Research Symposium EB2015. Journal of Investigative Dermatology, 2016, 136, 352-358.	0.7	63
117	Safety and early efficacy outcomes for lentiviral fibroblast gene therapy in recessive dystrophic epidermolysis bullosa. JCI Insight, 2019, 4, .	5.0	63
118	Detection of sequence variants in the gene encoding the Î²3 chain of laminin 5 (LAMB3). Human Mutation, 1995, 6, 77-84.	2.8	62
119	Quantitative Trait Loci for Yield under Multiple Stress and Drought Conditions in a Dry Bean Population. Crop Science, 2015, 55, 1596-1607.	1.9	62
120	The potential role of the gut microbiota in shaping host energetics and metabolic rate. Journal of Animal Ecology, 2020, 89, 2415-2426.	2.9	62
121	Use of laparoscopic videos amongst surgical trainees in the United Kingdom. Journal of the Royal College of Surgeons of Edinburgh, 2019, 17, 334-339.	2.0	61
122	Inherited disorders of desmosomes. Australasian Journal of Dermatology, 2005, 46, 221-229.	0.8	60
123	Detection of human papillomavirus type 18 E6 and E7-specific CD4+ T-helper 1 immunity in relation to health versus disease. International Journal of Cancer, 2006, 118, 950-956.	5.4	60
124	Genotypeâ€“Phenotype Correlation in Italian Patients with Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2002, 119, 1456-1462.	0.7	59
125	Impact of next generation sequencing on diagnostics in a genetic skin disease clinic. Experimental Dermatology, 2013, 22, 825-831.	2.9	59
126	Enhanced liver fibrosis test predicts transplantâ€free survival in primary sclerosing cholangitis, a multiâ€centre study. Liver International, 2017, 37, 1554-1561.	4.0	59

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127	Novel Molecular Therapies for Heritable Skin Disorders. Journal of Investigative Dermatology, 2012, 132, 820-828.	0.7	57
128	Phase I/II open-label trial of intravenous allogeneic mesenchymal stromal cell therapy in adults with recessive dystrophic epidermolysis bullosa. Journal of the American Academy of Dermatology, 2020, 83, 447-454.	1.2	57
129	A prospective prostate cancer screening programme for men with pathogenic variants in mismatch repair genes (IMPACT): initial results from an international prospective study. Lancet Oncology, The, 2021, 22, 1618-1631.	10.8	57
130	Cloning and developmental expression of pea ribulose-1,5-bisphosphate carboxylase/oxygenase large subunit N-methyltransferase. Plant Molecular Biology, 1995, 27, 249-261.	4.0	56
131	A Homozygous Nonsense Mutation in the Î±3 Chain Gene of Laminin 5 (LAMA3) in Herlitz Junctional Epidermolysis Bullosa: Prenatal Exclusion in a Fetus at Risk. Genomics, 1995, 29, 282-284.	2.9	56
132	Alterations in Desmosome Size and Number Coincide with the Loss of Keratinocyte Cohesion in Skin with Homozygous and Heterozygous Defects in the Desmosomal Protein Plakophilin 1. Journal of Investigative Dermatology, 2003, 121, 96-103.	0.7	56
133	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	55
134	Revertant Mosaicism in Recessive Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2010, 130, 1937-1940.	0.7	55
135	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	55
136	Schizophrenia: Gender and familial risk. Journal of Psychiatric Research, 1992, 26, 17-27.	3.2	54
137	Hemidesmosomes Show Abnormal Association with the Keratin Filament Network in Junctional Forms of Epidermolysis Bullosa. Journal of Investigative Dermatology, 1998, 110, 132-137.	0.7	54
138	Identification of the Active Site of Poly(A)-specific Ribonuclease by Site-directed Mutagenesis and Fe2+-mediated Cleavage. Journal of Biological Chemistry, 2002, 277, 5982-5987.	3.5	53
139	A novel translation re-initiation mechanism for the p63 gene revealed by amino-terminal truncating mutations in Rapp-Hodgkin/Hay-Wells-like syndromes. Human Molecular Genetics, 2008, 17, 1968-1977.	3.0	53
140	Clinicopathological Correlations of Compound Heterozygous COL7A1 Mutations in Recessive Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 1996, 107, 171-177.	0.7	52
141	Recurrent Mutations in the Type VII Collagen Gene (COL7A1) in Patients with Recessive Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 1997, 109, 246-249.	0.7	52
142	Novel IL31RA gene mutation and ancestral OSMR mutant allele in familial primary cutaneous amyloidosis. European Journal of Human Genetics, 2010, 18, 26-32.	2.9	52
143	Evolution of the <scp>S</scp>outhampton <scp>E</scp>nhanced <scp>R</scp>ecovery <scp>P</scp>rogramme for radical cystectomy and the aggregation of marginal gains. BJU International, 2014, 114, 375-383.	2.8	52
144	Inherited epidermolysis bullosa comes into the new millenium: A revised classification system based on current knowledge of pathogenetic mechanisms and the clinical, laboratory, and epidemiologic findings of large, well-defined patient cohorts. Journal of the American Academy of Dermatology, 2000, 43, 135-137.	1.2	51

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145	Parastomal hernia following cystectomy and ileal conduit urinary diversion: a systematic review. Hernia: the Journal of Hernias and Abdominal Wall Surgery, 2017, 21, 163-175.	2.1	51
146	Influence of the Second COL7A1 Mutation in Determining the Phenotypic Severity of Recessive Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 1996, 106, 766-770.	0.7	50
147	The Gene for Hypotrichosis of Marie Unna Maps between D8S258 and D8S298: Exclusion of the hr Gene by cDNA and Genomic Sequencing. American Journal of Human Genetics, 1999, 65, 413-419.	6.1	50
148	Patients with Recessive Dystrophic Epidermolysis Bullosa Develop Squamous-Cell Carcinoma Regardless of Type VII Collagen Expression. Journal of Investigative Dermatology, 2007, 127, 2438-2444.	0.7	49
149	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
150	Recessive mutation in tetraspanin CD151 causes Kindler syndrome-like epidermolysis bullosa with multi-systemic manifestations including nephropathy. Matrix Biology, 2018, 66, 22-33.	3.7	49
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