John A Mcgrath

List of Publications by Citations

650 papers	24,125 citations	74 h-index	124 g-index
694 ext. papers	27,725 ext. citations	4.5 avg, IF	6.63 L-index

#	Paper	IF	Citations
650	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, 931-50	4.5	690
649	Accumulation of Krebs cycle intermediates and over-expression of HIF1alpha in tumours which result from germline FH and SDH mutations. *Human Molecular Genetics*, **2005**, 14, 2231-9	5.6	683
648	Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. *Journal of the American Academy of Dermatology*, **2014**, 70, 1103-26	4.5	596
647	Alopecia universalis associated with a mutation in the human hairless gene. *Science*, **1998**, 279, 720-4	33.3	374
646	Prediction of real-world functional disability in chronic mental disorders: a comparison of schizophrenia and bipolar disorder. *American Journal of Psychiatry*, **2010**, 167, 1116-24	11.9	336
645	Revised classification system for inherited epidermolysis bullosa. *Journal of the American Academy of Dermatology*, **2000**, 42, 1051-1066	4.5	333
644	Mutations in the plakophilin 1 gene result in ectodermal dysplasia/skin fragility syndrome. *Nature Genetics*, **1997**, 17, 240-4	36.3	330
643	Bipolar I disorder and schizophrenia: a 440-single-nucleotide polymorphism screen of 64 candidate genes among Ashkenazi Jewish case-parent trios. *American Journal of Human Genetics*, **2005**, 77, 918-36	11	323
642	Mutations in the 180-kD bullous pemphigoid antigen (BPAG2), a hemidesmosomal transmembrane collagen (COL17A1), in generalized atrophic benign epidermolysis bullosa. *Nature Genetics*, **1995**, 11, 83-6	36.3	317
641	Hay-Wells syndrome is caused by heterozygous missense mutations in the SAM domain of p63. *Human Molecular Genetics*, **2001**, 10, 221-9	5.6	268
640	Loss of kindlin-1, a human homolog of the Caenorhabditis elegans actin-extracellular-matrix linker protein UNC-112, causes Kindler syndrome. *American Journal of Human Genetics*, **2003**, 73, 174-87	11	263
639	Bone marrow transplantation for recessive dystrophic epidermolysis bullosa. *New England Journal of Medicine*, **2010**, 363, 629-39	59.2	262
638	Loss of plectin causes epidermolysis bullosa with muscular dystrophy: cDNA cloning and genomic organization. *Genes and Development*, **1996**, 10, 1724-35	12.6	238
637	Autoantibodies to extracellular matrix protein 1 in lichen sclerosus. *Lancet, The*, **2003**, 362, 118-23	40	236
636	Germline NLRP1 Mutations Cause Skin Inflammatory and Cancer Susceptibility Syndromes via Inflammasome Activation. *Cell*, **2016**, 167, 187-202.e17	56.2	224
635	The role of fibroblasts in tissue engineering and regeneration. *British Journal of Dermatology*, **2007**, 156, 1149-55	4	218
634	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. *Nature Genetics*, **2013**, 45, 1244-1248	36.3	217

633	A molecular defect in loricrin, the major component of the cornified cell envelope, underlies Vohwinkel's syndrome. *Nature Genetics*, **1996**, 13, 70-7	36.3	214
632	Lipoid proteinosis maps to 1q21 and is caused by mutations in the extracellular matrix protein 1 gene (ECM1). *Human Molecular Genetics*, **2002**, 11, 833-40	5.6	192
631	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	11	180
630	Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *British Journal of Dermatology*, **2020**, 183, 614-627	4	179
629	Multicenter assessment of neoadjuvant chemotherapy for muscle-invasive bladder cancer. *European Urology*, **2015**, 67, 241-9	10.2	178
628	PDGFRalpha-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, 6609-14	11.5	178
627	Potential of fibroblast cell therapy for recessive dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **2008**, 128, 2179-89	4.3	176
626	The filaggrin story: novel insights into skin-barrier function and disease. *Trends in Molecular Medicine*, **2008**, 14, 20-7	11.5	174
625	Microdeletions of 3q29 confer high risk for schizophrenia. *American Journal of Human Genetics*, **2010**, 87, 229-36	11	173
624	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, 959-68	4.3	173
623	Targeted prostate cancer screening in BRCA1 and BRCA2 mutation carriers: results from the initial screening round of the IMPACT study. *European Urology*, **2014**, 66, 489-99	10.2	156
622	A homozygous nonsense mutation in the alpha 3 chain gene of laminin 5 (LAMA3) in lethal (Herlitz) junctional epidermolysis bullosa. *Human Molecular Genetics*, **1995**, 4, 959-62	5.6	156
621	Introduction of an enhanced recovery protocol for radical cystectomy. *BJU International*, **2008**, 101, 698-701	3.6	148
620	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		138
619	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, 581-9	11	137
618	Genome-wide association study of schizophrenia in Ashkenazi Jews. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168, 649-59	3.5	128
617	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, 360-6	4.3	123
616	The molecular basis of lipoid proteinosis: mutations in extracellular matrix protein 1. *Experimental Dermatology*, **2007**, 16, 881-90	4	117

615	Genetic diseases of junctions. *Journal of Investigative Dermatology*, **2007**, 127, 2713-25	4.3	113
614	Altered laminin 5 expression due to mutations in the gene encoding the beta 3 chain (LAMB3) in generalized atrophic benign epidermolysis bullosa. *Journal of Investigative Dermatology*, **1995**, 104, 467-74	4.3	113
613	Striate palmoplantar keratoderma resulting from desmoplakin haploinsufficiency. *Journal of Investigative Dermatology*, **1999**, 113, 940-6	4.3	112
612	Epidermolysis bullosa complicated by squamous cell carcinoma: report of 10 cases. *Journal of Cutaneous Pathology*, **1992**, 19, 116-23	1.7	110
611	Compound heterozygosity for non-sense and mis-sense mutations in desmoplakin underlies skin fragility/woolly hair syndrome. *Journal of Investigative Dermatology*, **2002**, 118, 232-8	4.3	109
610	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	4.3	109
609	Structural variations in anchoring fibrils in dystrophic epidermolysis bullosa: correlation with type VII collagen expression. *Journal of Investigative Dermatology*, **1993**, 100, 366-72	4.3	105
608	Kindlin-1 controls Wnt and TGF- availability to regulate cutaneous stem cell proliferation. *Nature Medicine*, **2014**, 20, 350-9	50.5	101
607	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, 1551-7	4.3	101
606	Epidermolysis bullosa pruriginosa: dystrophic epidermolysis bullosa with distinctive clinicopathological features. *British Journal of Dermatology*, **1994**, 130, 617-25	4	101
605	An unusual N-terminal deletion of the laminin alpha3a isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. *Human Molecular Genetics*, **2003**, 12, 2395-409	5.6	100
604	Oncostatin M receptor-beta mutations underlie familial primary localized cutaneous amyloidosis. *American Journal of Human Genetics*, **2008**, 82, 73-80	11	99
603	Relationship of the Brief UCSD Performance-based Skills Assessment (UPSA-B) to multiple indicators of functioning in people with schizophrenia and bipolar disorder. *Bipolar Disorders*, **2010**, 12, 45-55	3.8	95
602	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, 724-30	11	94
601	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, 601-11	11	94
600	Extracellular matrix protein 1 gene (ECM1) mutations in lipoid proteinosis and genotype-phenotype correlation. *Journal of Investigative Dermatology*, **2003**, 120, 345-50	4.3	93
599	Fibroblast cell therapy enhances initial healing in recessive dystrophic epidermolysis bullosa wounds: results of a randomized, vehicle-controlled trial. *British Journal of Dermatology*, **2013**, 169, 1025-33	4	92
598	Skin fragility and hypohidrotic ectodermal dysplasia resulting from ablation of plakophilin 1. *British Journal of Dermatology*, **1999**, 140, 297-307	4	92

597	Spectrum of dominant mutations in the desmosomal cadherin desmoglein 1, causing the skin disease striate palmoplantar keratoderma. *European Journal of Human Genetics*, **2001**, 9, 197-203	5.3	91
596	Mutational hotspots in the LAMB3 gene in the lethal (Herlitz) type of junctional epidermolysis bullosa. *Human Molecular Genetics*, **1996**, 5, 231-7	5.6	91
595	Revertant mosaicism in skin: natural gene therapy. *Trends in Molecular Medicine*, **2011**, 17, 140-8	11.5	90
594	Enhanced Recovery After Robot-assisted Radical Cystectomy: EAU Robotic Urology Section Scientific Working Group Consensus View. *European Urology*, **2016**, 70, 649-660	10.2	90
593	Potential of Systemic Allogeneic Mesenchymal Stromal Cell Therapy for Children with Recessive Dystrophic Epidermolysis Bullosa. *Journal of Investigative Dermatology*, **2015**, 135, 2319-2321	4.3	89
592	Lack of plakophilin 1 increases keratinocyte migration and reduces desmosome stability. *Journal of Cell Science*, **2003**, 116, 3303-14	5.3	89
591	3D In vitro model of a functional epidermal permeability barrier from human embryonic stem cells and induced pluripotent stem cells. *Stem Cell Reports*, **2014**, 2, 675-89	8	86
590	Cloning of the human type XVII collagen gene (COL17A1), and detection of novel mutations in generalized atrophic benign epidermolysis bullosa. *American Journal of Human Genetics*, **1997**, 60, 352-65	11	85
589	Fibroblast-derived dermal matrix drives development of aggressive cutaneous squamous cell carcinoma in patients with recessive dystrophic epidermolysis bullosa. *Cancer Research*, **2012**, 72, 3522-34	10.1	83
588	Genomewide linkage scan for bipolar-disorder susceptibility loci among Ashkenazi Jewish families. *American Journal of Human Genetics*, **2004**, 75, 204-19	11	83
587	Frameshift mutation in the V2 domain of human keratin 1 results in striate palmoplantar keratoderma. *Journal of Investigative Dermatology*, **2002**, 118, 838-44	4.3	83
586	Compound heterozygosity for a dominant glycine substitution and a recessive internal duplication mutation in the type XVII collagen gene results in junctional epidermolysis bullosa and abnormal dentition. *American Journal of Pathology*, **1996**, 148, 1787-96	5.8	83
585	Increased invasive behaviour in cutaneous squamous cell carcinoma with loss of basement-membrane type VII collagen. *Journal of Cell Science*, **2009**, 122, 1788-99	5.3	81
584	Kindler syndrome: a focal adhesion genodermatosis. *British Journal of Dermatology*, **2009**, 160, 233-42	4	81
583	Diseases of epidermal keratins and their linker proteins. *Experimental Cell Research*, **2007**, 313, 1995-2009	4.2	81
582	Clinical and molecular characterization of lipoid proteinosis in Namaqualand, South Africa. *British Journal of Dermatology*, **2004**, 151, 413-23	4	79
581	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. *European Urology*, **2019**, 76, 831-842	10.2	78
580	Fine mapping on chromosome 10q22-q23 implicates Neuregulin 3 in schizophrenia. *American Journal of Human Genetics*, **2009**, 84, 21-34	11	78

579	92 Reshaping the Diagnostic Pathways for Investigation of Haematuria During and After The COVID-19 Pandemic: Diagnostic Accuracy of Strategies for Detection of Bladder Cancer from The IDENTIFY Cohort Study. *British Journal of Surgery*, **2021**, 108,	5.3	78
578	Glycine substitutions in the triple-helical region of type VII collagen result in a spectrum of dystrophic epidermolysis bullosa phenotypes and patterns of inheritance. *American Journal of Human Genetics*, **1996**, 58, 671-81	11	76
577	Perioperative fluid management: Consensus statement from the enhanced recovery partnership. *Perioperative Medicine (London, England)*, **2012**, 1, 2	2.8	75
576	Infection and inflammation in schizophrenia and bipolar disorder: a genome wide study for interactions with genetic variation. *PLoS ONE*, **2015**, 10, e0116696	3.7	73
575	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	4.5	72
574	Management of cutaneous squamous cell carcinoma in patients with epidermolysis bullosa: best clinical practice guidelines. *British Journal of Dermatology*, **2016**, 174, 56-67	4	70
573	Colocalization of kindlin-1, kindlin-2, and migfilin at keratinocyte focal adhesion and relevance to the pathophysiology of Kindler syndrome. *Journal of Investigative Dermatology*, **2008**, 128, 2156-65	4.3	69
572	Sensitivity and specificity of the UCSD Performance-based Skills Assessment (UPSA-B) for identifying functional milestones in schizophrenia. *Schizophrenia Research*, **2011**, 132, 165-70	3.6	68
571	Extracellular matrix protein 1 inhibits the activity of matrix metalloproteinase 9 through high-affinity protein/protein interactions. *Experimental Dermatology*, **2006**, 15, 300-7	4	67
570	HPV-based cervical cancer screening in a population at high risk for HIV infection. *International Journal of Cancer*, **2000**, 85, 206-210	7.5	67
569	The role of kindlins in cell biology and relevance to human disease. *International Journal of Biochemistry and Cell Biology*, **2010**, 42, 595-603	5.6	65
568	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, 984-91	11	65
567	Molecular basis of recessive dystrophic epidermolysis bullosa: genotype/phenotype correlation in a case of moderate clinical severity. *Journal of Investigative Dermatology*, **1996**, 106, 119-24	4.3	65
566	Prenatal diagnosis for severe inherited skin disorders: 25 years' experience. *British Journal of Dermatology*, **2006**, 154, 106-13	4	64
565	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, 368-74	4.3	64
564	Epidermolysis bullosa. *Nature Reviews Disease Primers*, **2020**, 6, 78	51.1	64
563	Patient-specific naturally gene-reverted induced pluripotent stem cells in recessive dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **2014**, 134, 1246-1254	4.3	63
562	Limbal stem cell deficiency and ocular phenotype in ectrodactyly-ectodermal dysplasia-clefting syndrome caused by p63 mutations. *Ophthalmology*, **2012**, 119, 74-83	7.3	63

561	The international dystrophic epidermolysis bullosa patient registry: an online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. *Human Mutation*, **2011**, 32, 1100-7	4.7	63
560	Comparative mutation detection screening of the type VII collagen gene (COL7A1) using the protein truncation test, fluorescent chemical cleavage of mismatch, and conformation sensitive gel electrophoresis. *Journal of Investigative Dermatology*, **1999**, 113, 673-86	4.3	63
559	Familiality of novel factorial dimensions of schizophrenia. *Archives of General Psychiatry*, **2009**, 66, 591-600		61
558	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	4.3	61
557	Adjunctive testing for cervical cancer in low resource settings with visual inspection, HPV, and the Pap smear. *International Journal of Gynecology and Obstetrics*, **2001**, 72, 47-53	4	61
556	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, 771-4	4.3	60
555	Desmosomal genodermatoses. *British Journal of Dermatology*, **2012**, 166, 36-45	4	59
554	Allelic heterogeneity of dominant and recessive COL7A1 mutations underlying epidermolysis bullosa pruriginosa. *Journal of Investigative Dermatology*, **1999**, 112, 984-7	4.3	59
553	Schizophrenia: age at onset, gender and familial risk. *Acta Psychiatrica Scandinavica*, **1990**, 82, 344-51	6.5	59
552	WNT10A mutation causes ectodermal dysplasia by impairing progenitor cell proliferation and KLF4-mediated differentiation. *Nature Communications*, **2017**, 8, 15397	17.4	58
551	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	4.3	58
550	Heterozygous mutation in the SAM domain of p63 underlies Rapp-Hodgkin ectodermal dysplasia. *Journal of Dental Research*, **2003**, 82, 433-7	8.1	58
549	Corneodesmosin expression in psoriasis vulgaris differs from normal skin and other inflammatory skin disorders. *Laboratory Investigation*, **2001**, 81, 969-76	5.9	58
548	Detection of sequence variants in the gene encoding the beta 3 chain of laminin 5 (LAMB3). *Human Mutation*, **1995**, 6, 77-84	4.7	58
547	Progress in epidermolysis bullosa research: toward treatment and cure. *Journal of Investigative Dermatology*, **2010**, 130, 1778-84	4.3	57
546	Inherited disorders of desmosomes. *Australasian Journal of Dermatology*, **2005**, 46, 221-9	1.3	57
545	A comparison of different lysis buffers to assess allele dropout from single cells for preimplantation genetic diagnosis. *Prenatal Diagnosis*, **2001**, 21, 490-7	3.2	57
544	Frontal fibrosing alopecia: reflections and hypotheses on aetiology and pathogenesis. *Experimental Dermatology*, **2016**, 25, 847-852	4	57

543	Radiofrequency-induced Thermo-chemotherapy Effect Versus a Second Course of Bacillus Calmette-Guérin or Institutional Standard in Patients with Recurrence of Non-muscle-invasive Bladder Cancer Following Induction or Maintenance Bacillus Calmette-Guérin Therapy (HYMN): A Phase III, Open-label, Randomised Controlled Trial. *European Urology*, **2019**, 75, 63-71	10.2	57
542	Novel and emerging therapies in the treatment of recessive dystrophic epidermolysis bullosa. *Intractable and Rare Diseases Research*, **2017**, 6, 6-20	1.4	56
541	Kindler syndrome. *Dermatologic Clinics*, **2010**, 28, 119-24	4.2	56
540	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. *Nature Genetics*, **2012**, 44, 1272-6	36.3	56
539	Genome-wide association study in frontal fibrosing alopecia identifies four susceptibility loci including HLA-B*07:02. *Nature Communications*, **2019**, 10, 1150	17.4	55
538	Recurrent heterozygous missense mutation, p.Gly573Ser, in the TRPV3 gene in an Indian boy with sporadic Olmsted syndrome. *British Journal of Dermatology*, **2012**, 167, 440-2	4	55
537	Keratin 9 is required for the structural integrity and terminal differentiation of the palmoplantar epidermis. *Journal of Investigative Dermatology*, **2014**, 134, 754-763	4.3	55
536	Ectodermal dysplasia-skin fragility syndrome. *Dermatologic Clinics*, **2010**, 28, 125-9	4.2	55
535	An essential role for the Zn transporter ZIP7 in B cell development. *Nature Immunology*, **2019**, 20, 350-361	19.1	54
534	Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. *British Journal of Dermatology*, **2015**, 172, 94-100	4	54
533	Reciprocal duplication of the Williams-Beuren syndrome deletion on chromosome 7q11.23 is associated with schizophrenia. *Biological Psychiatry*, **2014**, 75, 371-7	7.9	54
532	Integrative mRNA profiling comparing cultured primary cells with clinical samples reveals PLK1 and C20orf20 as therapeutic targets in cutaneous squamous cell carcinoma. *Oncogene*, **2011**, 30, 4666-77	9.2	54
531	Matrix metalloproteinase-7 activates heparin-binding epidermal growth factor-like growth factor in cutaneous squamous cell carcinoma. *British Journal of Dermatology*, **2010**, 163, 726-35	4	54
530	HB-EGF induces COL7A1 expression in keratinocytes and fibroblasts: possible mechanism underlying allogeneic fibroblast therapy in recessive dystrophic epidermolysis Bullosa. *Journal of Investigative Dermatology*, **2011**, 131, 1771-4	4.3	54
529	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	3	54
528	Lichen planus and lichenoid dermatoses: Clinical overview and molecular basis. *Journal of the American Academy of Dermatology*, **2018**, 79, 789-804	4.5	54
527	APOBEC mutation drives early-onset squamous cell carcinomas in recessive dystrophic epidermolysis bullosa. *Science Translational Medicine*, **2018**, 10,	17.5	53
526	Association of obesity and treated hypertension and diabetes with cognitive ability in bipolar disorder and schizophrenia. *Bipolar Disorders*, **2014**, 16, 422-31	3.8	53

525	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, 314-21	4.3	53
524	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	4.3	52
523	Transplanted bone marrow-derived circulating PDGFR□ cells restore type VII collagen in recessive dystrophic epidermolysis bullosa mouse skin graft. *Journal of Immunology*, **2015**, 194, 1996-2003	5.3	52
522	Characterization of IgG autoantibodies to extracellular matrix protein 1 in lichen sclerosus. *Clinical and Experimental Dermatology*, **2004**, 29, 499-504	1.8	52
521	Alpha 6 beta 4 integrin abnormalities in junctional epidermolysis bullosa with pyloric atresia. *British Journal of Dermatology*, **2001**, 144, 408-14	4	52
520	LAP-VEGaS Practice Guidelines for Reporting of Educational Videos in Laparoscopic Surgery: A Joint Trainers and Trainees Consensus Statement. *Annals of Surgery*, **2018**, 268, 920-926	7.8	51
519	Epithelial inflammation resulting from an inherited loss-of-function mutation in EGFR. *Journal of Investigative Dermatology*, **2014**, 134, 2570-2578	4.3	51
518	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	4.3	51
517	New insight into mechanisms of pruritus from molecular studies on familial primary localized cutaneous amyloidosis. *British Journal of Dermatology*, **2009**, 161, 1217-24	4	50
516	Hemidesmosomes show abnormal association with the keratin filament network in junctional forms of epidermolysis bullosa. *Journal of Investigative Dermatology*, **1998**, 110, 132-7	4.3	50
515	Striate palmoplantar keratoderma arising from desmoplakin and desmoglein 1 mutations is associated with contrasting perturbations of desmosomes and the keratin filament network. *British Journal of Dermatology*, **2004**, 150, 878-91	4	50
514	Evaluation of a human papillomavirus assay in cervical screening in Zimbabwe. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2000**, 107, 33-8	3.7	50
513	A homozygous nonsense mutation in the alpha 3 chain gene of laminin 5 (LAMA3) in Herlitz junctional epidermolysis bullosa: prenatal exclusion in a fetus at risk. *Genomics*, **1995**, 29, 282-4	4.3	50
512	Epidermolysis bullosa simplex (Dowling-Meara). A clinicopathological review. *British Journal of Dermatology*, **1992**, 126, 421-30	4	50
511	Germline mutation in ATR in autosomal- dominant oropharyngeal cancer syndrome. *American Journal of Human Genetics*, **2012**, 90, 511-7	11	49
510	Novel molecular therapies for heritable skin disorders. *Journal of Investigative Dermatology*, **2012**, 132, 820-8	4.3	49
509	Genotype-phenotype correlation in skin fragility-ectodermal dysplasia syndrome resulting from mutations in plakophilin 1. *Experimental Dermatology*, **2002**, 11, 107-14	4	49
508	Genotype-phenotype correlation in italian patients with dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **2002**, 119, 1456-62	4.3	49

507	Progress toward Treatment and Cure of Epidermolysis Bullosa: Summary of the DEBRA International Research Symposium EB2015. *Journal of Investigative Dermatology*, **2016**, 136, 352-358	4.3	48
506	Suppression of TGF and Angiogenesis by Type VII Collagen in Cutaneous SCC. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	48
505	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. *American Journal of Human Genetics*, **2012**, 91, 1115-21	11	48
504	Trends in operative caseload and mortality rates after radical cystectomy for bladder cancer in England for 1998-2010. *European Urology*, **2015**, 67, 1056-1062	10.2	47
503	Revertant mosaicism in recessive dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **2010**, 130, 1937-40	4.3	47
502	Recurrent mutations in the type VII collagen gene (COL7A1) in patients with recessive dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **1997**, 109, 246-9	4.3	47
501	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, 1776-83	4.3	47
500	Risk factors in schizophrenia. Season of birth, gender, and familial risk. *British Journal of Psychiatry*, **1992**, 160, 65-71	5.4	47
499	Oral and gastrointestinal manifestations of epidermolysis bullosa. *Lancet, The*, **1992**, 340, 1505-6	40	47
498	Transformation-specific matrix metalloproteinases (MMP)-7 and MMP-13 are expressed by tumour cells in epidermolysis bullosa-associated squamous cell carcinomas. *British Journal of Dermatology*, **2008**, 158, 778-85	4	46
497	Schizophrenia: gender and familial risk. *Journal of Psychiatric Research*, **1992**, 26, 17-27	5.2	46
496	MUTATION-BASED PRENATAL DIAGNOSIS OF HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA **1997**, 17, 343-354		45
495	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, 413-9	11	45
494	Clinicopathological correlations of compound heterozygous COL7A1 mutations in recessive dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **1996**, 107, 171-7	4.3	45
493	Thalidomide in the management of epidermolysis bullosa pruriginosa. *British Journal of Dermatology*, **2005**, 152, 1332-4	4	44
492	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, 135-7	4.5	44
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