

# John A Mcgrath

## List of Publications by Year in Descending Order

**Source:** <https://exaly.com/author-pdf/2115638/john-a-mcgrath-publications-by-year.pdf>

**Version:** 2024-04-28

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	P63 targeted deletion under the FOXP1 promoter disrupts pre-and post-natal thymus development, function and maintenance as well as induces severe hair loss.. <i>PLoS ONE</i> , <b>2022</b> , 17, e0261770	2.7	0
649	Transcriptomic response of peripheral blood mononuclear cells to secukinumab in an 8-year-old boy with juvenile generalized pustular psoriasis.. <i>Journal of the European Academy of Dermatology and Venereology</i> , <b>2022</b> ,	4.6	
648	Identifying the Optimal Number of Neoadjuvant Chemotherapy Cycles in Patients with Muscle Invasive Bladder Cancer. <i>Journal of Urology</i> , <b>2022</b> , 207, 70-76	2.5	1
647	WNT10A variant and severe scoliosis?. <i>Journal of Dermatology</i> , <b>2022</b> ,	1.6	0
646	Incontinentia pigmenti in a male infant and a proposed diagnostic algorithm.. <i>Clinical and Experimental Dermatology</i> , <b>2022</b> ,	1.8	
645	Metabolic syndrome and comorbidities in patients with psoriasis: a community-based case-control study from the Nagahama cohort in Japan.. <i>European Journal of Dermatology</i> , <b>2022</b> , 32, 86-93	0.8	1
644	Autosomal dominant epidermolysis bullosa simplex exacerbated by hyperkeratotic scabies.. <i>Journal of Dermatology</i> , <b>2022</b> ,	1.6	
643	Restoring type VII collagen in skin.. <i>Med</i> , <b>2022</b> , 3, 273-275	31.7	
642	4/9 Integrins Coordinate Epithelial Cell Migration Through Local Suppression of MAP Kinase Signaling Pathways.. <i>Frontiers in Cell and Developmental Biology</i> , <b>2021</b> , 9, 750771	5.7	1
641	The potential of gene therapy for recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2021</b> ,	4	1
640	Autoinflammatory Keratinization Disease With Hepatitis and Autism Reveals Roles for JAK1 Kinase Hyperactivity in Autoinflammation.. <i>Frontiers in Immunology</i> , <b>2021</b> , 12, 737747	8.4	0
639	A prospective prostate cancer screening programme for men with pathogenic variants in mismatch repair genes (IMPACT): initial results from an international prospective study. <i>Lancet Oncology</i> , <b>2021</b> , 22, 1618-1631	21.7	10
638	Plasma metabolomic and lipidomic profiling highlights metabolic changes in keloid-prone individuals. <i>Experimental Dermatology</i> , <b>2021</b> ,	4	0
637	Neoadjuvant chemotherapy plus radical cystectomy versus radical cystectomy alone in clinical T2 bladder cancer without hydronephrosis. <i>BJU International</i> , <b>2021</b> , 128, 79-87	5.6	7
636	A de novo COL17A1 splice-site mutation causing a 7-bp deletion in a Taiwanese patient with junctional epidermolysis bullosa. <i>European Journal of Dermatology</i> , <b>2021</b> , 31, 267-269	0.8	
635	Molecular epidemiology of non-syndromic autosomal recessive congenital ichthyosis in a Middle-Eastern population. <i>Experimental Dermatology</i> , <b>2021</b> , 30, 1290-1297	4	3
634	How reliable are surgeon-reported data? A comparison of the British Association of Urological Surgeons radical prostatectomy audit with the National Prostate Cancer Audit Hospital Episode Statistics-linked database. <i>BJU International</i> , <b>2021</b> , 128, 482-489	5.6	4

633	Kindler epidermolysis bullosa-like skin phenotype and downregulated basement membrane zone gene expression in poikiloderma with neutropenia and a homozygous USB1 mutation. <i>Matrix Biology</i> , <b>2021</b> , 99, 43-57	11.4	0
632	Synergistic multiple early therapy (SMET) for inflammatory diseases with pathogenic autoinflammatory feedback circuits. <i>British Journal of Dermatology</i> , <b>2021</b> , 185, 469-470	4	
631	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. <i>British Journal of Surgery</i> , <b>2021</b> , 108,	5.3	78
630	A truncating variant in SERPINA3, skin pustules and adult-onset immunodeficiency. <i>Journal of Dermatology</i> , <b>2021</b> , 48, e370-e371	1.6	
629	The diagnostic impact of UK regional variations in age-specific prostate-specific antigen guidelines. <i>BJU International</i> , <b>2021</b> , 128, 298-300	5.6	2
628	There is no proven association between sensitization to benzyl salicylate and frontal fibrosing alopecia. <i>Contact Dermatitis</i> , <b>2021</b> , 85, 483-484	2.7	0
627	Investigational Treatments for Epidermolysis Bullosa. <i>American Journal of Clinical Dermatology</i> , <b>2021</b> , 22, 801-817	7.1	2
626	How to report educational videos in robotic surgery: an international multidisciplinary consensus statement. <i>Updates in Surgery</i> , <b>2021</b> , 73, 815-821	2.9	6
625	A decade of next-generation sequencing in genodermatoses: the impact on gene discovery and clinical diagnostics. <i>British Journal of Dermatology</i> , <b>2021</b> , 184, 606-616	4	3
624	Mutations in genes encoding desmosomal proteins: spectrum of cutaneous and extracutaneous abnormalities. <i>British Journal of Dermatology</i> , <b>2021</b> , 184, 596-605	4	8
623	Autosomal recessive hypotrichosis with loose anagen hairs associated with TKFC mutations. <i>British Journal of Dermatology</i> , <b>2021</b> , 184, 935-943	4	2
622	Heterogeneous addiction to transforming growth factor-beta signalling in recessive dystrophic epidermolysis bullosa-associated cutaneous squamous cell carcinoma. <i>British Journal of Dermatology</i> , <b>2021</b> , 184, 697-708	4	3
621	Comparative effectiveness of neoadjuvant chemotherapy in bladder and upper urinary tract urothelial carcinoma. <i>BJU International</i> , <b>2021</b> , 127, 528-537	5.6	1
620	Nanomedicine Approaches to Negotiate Local Biobarriers for Topical Drug Delivery. <i>Advanced Therapeutics</i> , <b>2021</b> , 4, 2000160	4.9	1
619	A germline mutation in the platelet-derived growth factor receptor beta gene may be implicated in hereditary progressive mucinous histiocytosis. <i>British Journal of Dermatology</i> , <b>2021</b> , 184, 967-970	4	5
618	Slac2-b Coordinates Extracellular Vesicle Secretion to Regulate Keratinocyte Adhesion and Migration. <i>Journal of Investigative Dermatology</i> , <b>2021</b> , 141, 523-532.e2	4.3	1
617	Immune tolerance of allogeneic haematopoietic cell transplantation supports donor epidermal grafting of recessive dystrophic epidermolysis bullosa chronic wounds. <i>British Journal of Dermatology</i> , <b>2021</b> , 184, 1161-1169	4	5
616	Prevalence, pathophysiology and management of itch in epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2021</b> , 184, 816-825	4	12

615	Additional Treatments to the Local tumour for metastatic prostate cancer-Assessment of Novel Treatment Algorithms (IP2-ATLANTA): protocol for a multicentre, phase II randomised controlled trial. <i>BMJ Open</i> , <b>2021</b> , 11, e042953	3	6
614	Spatial activation of ezrin by epidermal growth factor receptor and focal adhesion kinase co-ordinates epithelial cell migration. <i>Open Biology</i> , <b>2021</b> , 11, 210166	7	0
613	Ancestral patterns of recessive dystrophic epidermolysis bullosa mutations in Hispanic populations suggest sephardic ancestry. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 3390-3400	2.5	
612	Association of age with response to preoperative chemotherapy in patients with muscle-invasive bladder cancer. <i>World Journal of Urology</i> , <b>2021</b> , 39, 4345-4354	4	2
611	Induced pluripotent stem cell (iPSC) line MLI-004A derived from a patient with recessive dystrophic epidermolysis bullosa (RDEB). <i>Stem Cell Research</i> , <b>2021</b> , 55, 102463	1.6	
610	SERPINA1, generalized pustular psoriasis, and adult-onset immunodeficiency. <i>Journal of Dermatology</i> , <b>2021</b> , 48, 1597-1601	1.6	0
609	WNT10A, dermatology and dentistry. <i>British Journal of Dermatology</i> , <b>2021</b> ,	4	7
608	The natural history of laryngo-onycho-cutaneous syndrome: A case series of six pediatric patients and literature review. <i>Pediatric Dermatology</i> , <b>2021</b> , 38, 1094-1101	1.9	4
607	Clinical characteristics of male frontal fibrosing alopecia: a single-centre case series from London, UK. <i>British Journal of Dermatology</i> , <b>2021</b> ,	4	
606	The IDENTIFY study: the investigation and detection of urological neoplasia in patients referred with suspected urinary tract cancer - a multicentre observational study. <i>BJU International</i> , <b>2021</b> , 128, 440-450	5.6	4
605	Metabolic perturbations in fibrosis disease. <i>International Journal of Biochemistry and Cell Biology</i> , <b>2021</b> , 139, 106073	5.6	0
604	Unravelling the genetic basis of contact allergy. <i>Contact Dermatitis</i> , <b>2021</b> ,	2.7	1
603	The epidemiology of epidermolysis bullosa in England and Wales: data from the national epidermolysis bullosa database.. <i>British Journal of Dermatology</i> , <b>2021</b> ,	4	1
602	Current topics in Epidermolysis bullosa: Pathophysiology and therapeutic challenges.. <i>Journal of Dermatological Science</i> , <b>2021</b> , 104, 164-176	4.3	0
601	Arrhythmogenic right ventricular cardiomyopathy in patients with biallelic JUP-associated skin fragility. <i>Scientific Reports</i> , <b>2020</b> , 10, 21622	4.9	3
600	New Homozygous Missense MSMO1 Mutation in Two Siblings with SC4MOL Deficiency Presenting with Psoriasiform Dermatitis. <i>Cytogenetic and Genome Research</i> , <b>2020</b> , 160, 523-530	1.9	1
599	Novel p.Ala675Thr missense mutation in TRPV3 in Olmsted syndrome. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 796-798	1.8	1
598	Promising effect of intravenous immunoglobulin therapy for epidermolysis bullosa pruriginosa. <i>International Journal of Dermatology</i> , <b>2020</b> , 59, 851-855	1.7	1

597	Alopecia areata and frontal fibrosing alopecia: dimorphism by concurrence. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 734-737	1.8	2
596	Autosomal recessive mutations in plakoglobin and risk of cardiac abnormalities. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 654-657	1.8	1
595	IDENTIFY: The investigation and detection of urological neoplasia in patients referred with suspected urinary tract cancer: A multicentre cohort study. <i>International Journal of Surgery Protocols</i> , <b>2020</b> , 21, 8-12	1.1	2
594	Diagnostic Assessment of Lower Urinary Tract Symptoms in Men Considering Prostate Surgery: A Noninferiority Randomised Controlled Trial of Urodynamics in 26 Hospitals. <i>European Urology</i> , <b>2020</b> , 78, 701-710	10.2	15
593	Hispanic/Latino heritage group disparities in sleep and the sleep-cardiovascular health relationship by housing tenure status in the United States. <i>Sleep Health</i> , <b>2020</b> , 6, 451-462	4	3
592	Critical analysis of quality of life and cost-effectiveness of enhanced recovery after surgery (ERAS) for patient's undergoing urologic oncology surgery: a systematic review. <i>World Journal of Urology</i> , <b>2020</b> , 1	4	9
591	Reporting Radical Cystectomy Outcomes Following Implementation of Enhanced Recovery After Surgery Protocols: A Systematic Review and Individual Patient Data Meta-analysis. <i>European Urology</i> , <b>2020</b> , 78, 719-730	10.2	18
590	Molecular basis and inheritance patterns of amyloidosis cutis dyschromica. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 650-653	1.8	5
589	Impact of sex on response to neoadjuvant chemotherapy in patients with bladder cancer. <i>Urologic Oncology: Seminars and Original Investigations</i> , <b>2020</b> , 38, 639.e1-639.e9	2.8	7
588	Genomics-based treatment in a patient with two overlapping heritable skin disorders: Epidermolysis bullosa and acrodermatitis enteropathica. <i>Human Mutation</i> , <b>2020</b> , 41, 906-912	4.7	7
587	Ectodermal dysplasia-skin fragility syndrome: Two new cases and review of this desmosomal genodermatosis. <i>Experimental Dermatology</i> , <b>2020</b> , 29, 520-530	4	1
586	Epidermolysis bullosa (EB) pruriginosa associated with recessive homozygous mutations in COL7A1: case report of a rare EB genotype-phenotype. <i>Journal of the European Academy of Dermatology and Venereology</i> , <b>2020</b> , 34, e501-e504	4.6	
585	Genetic analysis in three Egyptian patients with Griscelli syndrome Type 1 reveals new nonsense mutations in MYO5A. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 789-792	1.8	5
584	Detailed hair shaft analysis in a man with delayed-onset Chediak-Higashi syndrome. <i>British Journal of Dermatology</i> , <b>2020</b> , 182, 223-225	4	2
583	Initial experience of the adjuvant treatments to the local tumor for metastatic prostate cancer: Assessment of novel treatment algorithms, a multicenter, phase II randomized controlled trial (IP2-ATLANTA).. <i>Journal of Clinical Oncology</i> , <b>2020</b> , 38, TPS5600-TPS5600	2.2	1
582	Sleep Health and Serious Psychological Distress: A Nationally Representative Study of the United States among White, Black, and Hispanic/Latinx Adults. <i>Nature and Science of Sleep</i> , <b>2020</b> , 12, 1091-1104 <sup>3.6</sup>	3.6	10
581	Urodynamics tests for the diagnosis and management of bladder outlet obstruction in men: the UPSTREAM non-inferiority RCT. <i>Health Technology Assessment</i> , <b>2020</b> , 24, 1-122	4.4	4
580	Genetics of Scars and Keloids <b>2020</b> , 47-53		

579	Coagulation Factor XIII-A Subunit Missense Mutation in the Pathobiology of Autosomal Dominant Multiple Dermatofibromas. <i>Journal of Investigative Dermatology</i> , <b>2020</b> , 140, 624-635.e7	4.3	4
578	The prognostic value of the neutrophil-to-lymphocyte ratio in patients with muscle-invasive bladder cancer treated with neoadjuvant chemotherapy and radical cystectomy. <i>Urologic Oncology: Seminars and Original Investigations</i> , <b>2020</b> , 38, 3.e17-3.e27	2.8	15
577	Sneddon syndrome associated with two novel ADA2 gene mutations. <i>Rheumatology</i> , <b>2020</b> , 59, 1448-1450.9	5	5
576	EBGene trial: patient preselection outcomes for the European GENEGRAFT ex vivo phase I/II gene therapy trial for recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2020</b> , 182, 794-797	4	15
575	Schopf-Schulz-Passarge syndrome: a rare ectodermal dysplasia with a delayed diagnosis. <i>International Journal of Dermatology</i> , <b>2020</b> , 59, 257-258	1.7	1
574	GrisCELLI syndrome type 3 in Ethiopian sisters resulting from a homozygous missense mutation in MLPH. <i>International Journal of Dermatology</i> , <b>2020</b> , 59, e55-e57	1.7	2
573	Pseudoporphyria induced by ultraviolet radiation. <i>Australasian Journal of Dermatology</i> , <b>2020</b> , 61, 177-179.3	3	3
572	Phase I/II open-label trial of intravenous allogeneic mesenchymal stromal cell therapy in adults with recessive dystrophic epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , <b>2020</b> , 83, 447-454	4.5	26
571	Homozygous Nonsense Mutation in DSC3 Resulting in Skin Fragility and Hypotrichosis. <i>Journal of Investigative Dermatology</i> , <b>2020</b> , 140, 1285-1288	4.3	4
570	ESDR and the European Dermatology Forum: 'ESDR's Baby' Big Brother. <i>Journal of Investigative Dermatology</i> , <b>2020</b> , 140, S181-S182	4.3	3
569	Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. <i>British Journal of Dermatology</i> , <b>2020</b> , 183, 614-627	4	179
568	Frontal fibrosing alopecia: a descriptive cross-sectional study of 711 cases in female patients from the UK. <i>British Journal of Dermatology</i> , <b>2020</b> , 183, 1136-1138	4	5
567	Phenotypic suppression of acral peeling skin syndrome in a patient with autosomal recessive congenital ichthyosis. <i>Experimental Dermatology</i> , <b>2020</b> , 29, 742-748	4	0
566	Epidermolysis bullosa. <i>Nature Reviews Disease Primers</i> , <b>2020</b> , 6, 78	51.1	64
565	Blaschko-linear lichen planus: Clinicopathological and genetic analysis. <i>Journal of Dermatology</i> , <b>2020</b> , 47, e384-e385	1.6	0
564	PLACK syndrome: the penny dropped. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 1091-1092	1.8	3
563	Genotype-phenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. <i>British Journal of Dermatology</i> , <b>2020</b> , 182, 729-737	4	19
562	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 391-394	1.8	1

561	Human Mesenchymal Stromal Cells Engineered to Express Collagen VII Can Restore Anchoring Fibrils in Recessive Dystrophic Epidermolysis Bullosa Skin Graft Chimeras. <i>Journal of Investigative Dermatology</i> , <b>2020</b> , 140, 121-131.e6	4.3	9
560	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. <i>European Urology</i> , <b>2019</b> , 76, 831-842	10.2	78
559	Perioperative and oncological outcomes of radical prostatectomy for high-risk prostate cancer in the UK: an analysis of surgeon-reported data. <i>BJU International</i> , <b>2019</b> , 124, 441-448	5.6	6
558	Nail lichen planus. <i>Journal of the American Academy of Dermatology</i> , <b>2019</b> , 80, e179	4.5	2
557	An essential role for the Zn transporter ZIP7 in B cell development. <i>Nature Immunology</i> , <b>2019</b> , 20, 350-361	19.1	54
556	EMSY expression affects multiple components of the skin barrier with relevance to atopic dermatitis. <i>Journal of Allergy and Clinical Immunology</i> , <b>2019</b> , 144, 470-481	11.5	12
555	Bone marrow transplant with post-transplant cyclophosphamide for recessive dystrophic epidermolysis bullosa expands the related donor pool and permits tolerance of nonhaematopoietic cellular grafts. <i>British Journal of Dermatology</i> , <b>2019</b> , 181, 1238-1246	4	20
554	Identification of Rigosertib for the Treatment of Recessive Dystrophic Epidermolysis Bullosa-Associated Squamous Cell Carcinoma. <i>Clinical Cancer Research</i> , <b>2019</b> , 25, 3384-3391	12.9	16
553	Genetic analysis in Egyptian patients with Chediak-Higashi syndrome reveals new LYST mutations. <i>Clinical and Experimental Dermatology</i> , <b>2019</b> , 44, 814-817	1.8	1
552	Pterygium and thinning of nails as an unusual manifestation in Clouston syndrome. <i>Journal of Dermatology</i> , <b>2019</b> , 46, e329-e330	1.6	3
551	Genome-wide association study in frontal fibrosing alopecia identifies four susceptibility loci including HLA-B*07:02. <i>Nature Communications</i> , <b>2019</b> , 10, 1150	17.4	55
550	Thrombospondin-1 Is a Major Activator of TGF- $\beta$ Signaling in Recessive Dystrophic Epidermolysis Bullosa Fibroblasts. <i>Journal of Investigative Dermatology</i> , <b>2019</b> , 139, 1497-1505.e5	4.3	17
549	Frontal fibrosing alopecia should be renamed to lichen planopilaris of Kossard. <i>Journal of the American Academy of Dermatology</i> , <b>2019</b> , 81, e53	4.5	2
548	Dermatological manifestations of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis (POIKTMP): a case series of 28 patients. <i>British Journal of Dermatology</i> , <b>2019</b> , 181, 862-864	4	3
547	Revertant mosaic fibroblasts in recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2019</b> , 181, 1247-1253	4	15
546	Biallelic KRT5 mutations in autosomal recessive epidermolysis bullosa simplex, including a complete human keratin 5 "knock-out". <i>Matrix Biology</i> , <b>2019</b> , 83, 48-59	11.4	8
545	Generation and Clinical Application of Gene-Modified Autologous Epidermal Sheets in Netherton Syndrome: Lessons Learned from a Phase 1 Trial. <i>Human Gene Therapy</i> , <b>2019</b> , 30, 1067-1078	4.8	15
544	Clinical and Patient-reported Outcome Measures in Men Referred for Consideration of Surgery to Treat Lower Urinary Tract Symptoms: Baseline Results and Diagnostic Findings of the Urodynamics for Prostate Surgery Trial; Randomised Evaluation of Assessment Methods (UPSTREAM). <i>European Urology Focus</i> , <b>2019</b> , 5, 340-350	5.1	13

543	Semidominant GPNMB Mutations in Amyloidosis Cutis Dyschromica. <i>Journal of Investigative Dermatology</i> , <b>2019</b> , 139, 2550-2554.e9	4.3	7
542	Safety and early efficacy outcomes for lentiviral fibroblast gene therapy in recessive dystrophic epidermolysis bullosa. <i>JCI Insight</i> , <b>2019</b> , 4,	9.9	30
541	Multicomponent hospital-led interventions to reduce hospital stay for older adults following elective surgery: a systematic review. <i>Health Services and Delivery Research</i> , <b>2019</b> , 7, 1-178	1.5	4
540	Dr. John McGrath. <i>Nishinon Journal of Dermatology</i> , <b>2019</b> , 81, 427-428	0	
539	Photodynamic versus white light-guided treatment of non-muscle invasive bladder cancer: a study protocol for a randomised trial of clinical and cost-effectiveness. <i>BMJ Open</i> , <b>2019</b> , 9, e022268	3	8
538	Use of laparoscopic videos amongst surgical trainees in the United Kingdom. <i>Journal of the Royal College of Surgeons of Edinburgh</i> , <b>2019</b> , 17, 334-339	2.5	29
537	Mutations in PLOD3, encoding lysyl hydroxylase 3, cause a complex connective tissue disorder including recessive dystrophic epidermolysis bullosa-like blistering phenotype with abnormal anchoring fibrils and type VII collagen deficiency. <i>Matrix Biology</i> , <b>2019</b> , 81, 91-106	11.4	28
536	Consanguinity and Double Recessive Gene Pathology: Cutis Laxa (PYCR1) and Nephrotic Syndrome (PLCE1). <i>JAMA Dermatology</i> , <b>2019</b> , 155, 257-259	5.1	4
535	Kindlin-1 Regulates Epidermal Growth Factor Receptor Signaling. <i>Journal of Investigative Dermatology</i> , <b>2019</b> , 139, 369-379	4.3	6
534	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 Randomized Controlled Trial. <i>Journal of Clinical Oncology</i> , <b>2019</b> , 37, 63-71	10.2	57
533	Coinheritance of 2 New Potentially Damaging Heterozygous COL7A1 Variants in a Family With Autosomal Dominant Epidermolysis Bullosa Pruriginosa. <i>Pediatric and Developmental Pathology</i> , <b>2018</b> , 21, 580-584	2.2	
532	LAP-VEGaS Practice Guidelines for Reporting of Educational Videos in Laparoscopic Surgery: A Joint Trainers and Trainees Consensus Statement. <i>Annals of Surgery</i> , <b>2018</b> , 268, 920-926	7.8	51
531	A case report of squamous cell carcinoma in a suprapubic urinary catheter tract: surgical excision and simultaneous colostomy formation. <i>Journal of Surgical Case Reports</i> , <b>2018</b> , 2018, rjy030	0.6	1
530	Autoinflammatory keratinization diseases: An emerging concept encompassing various inflammatory keratinization disorders of the skin. <i>Journal of Dermatological Science</i> , <b>2018</b> , 90, 105-111	4.3	37
529	Lung Function in Oil Spill Response Workers 1-3 Years After the Deepwater Horizon Disaster. <i>Epidemiology</i> , <b>2018</b> , 29, 315-322	3.1	20
528	Open radical cystectomy in England: the current standard of care - an analysis of the British Association of Urological Surgeons (BAUS) cystectomy audit and Hospital Episodes Statistics (HES) data. <i>BJU International</i> , <b>2018</b> , 121, 880-885	5.6	11
527	EB2017-Progress in Epidermolysis Bullosa Research toward Treatment and Cure. <i>Journal of Investigative Dermatology</i> , <b>2018</b> , 138, 1010-1016	4.3	30
526	Concurrent hidradenitis suppurativa and Dowling-Degos disease taken down a 'Notch'. <i>British Journal of Dermatology</i> , <b>2018</b> , 178, 328	4	6

525	The British Association of Urological Surgeons (BAUS) radical prostatectomy audit 2014/2015 - an update on current practice and outcomes by centre and surgeon case-volume. <i>BJU International</i> , <b>2018</b> , 121, 886-892	5.6	22
524	Exposure to Oil Spill Chemicals and Lung Function in Deepwater Horizon Disaster Response Workers. <i>Journal of Occupational and Environmental Medicine</i> , <b>2018</b> , 60, e312-e318	2	9
523	Homozygous acceptor splice site mutation in DSG1 disrupts plakoglobin localization and results in keratoderma and skin fragility. <i>Journal of Dermatological Science</i> , <b>2018</b> , 89, 198-201	4.3	7
522	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. <i>British Journal of Cancer</i> , <b>2018</b> , 118, 266-276	8.7	9
521	HLA typing using genome wide data reveals susceptibility types for infections in a psychiatric disease enriched sample. <i>Brain, Behavior, and Immunity</i> , <b>2018</b> , 70, 203-213	16.6	6
520	Beneficial effect of ustekinumab in familial pityriasis rubra pilaris with a new missense mutation in CARD14. <i>British Journal of Dermatology</i> , <b>2018</b> , 178, 969-972	4	29
519	Inherited palmoplantar keratodermas: the heart of the matter. <i>Clinical and Experimental Dermatology</i> , <b>2018</b> , 43, 228-230	1.8	0
518	Caveolin-1 Controls Hyperresponsiveness to Mechanical Stimuli and Fibrogenesis-Associated RUNX2 Activation in Keloid Fibroblasts. <i>Journal of Investigative Dermatology</i> , <b>2018</b> , 138, 208-218	4.3	42
517	Time Series Integrative Analysis of RNA Sequencing and MicroRNA Expression Data Reveals Key Biologic Wound Healing Pathways in Keloid-Prone Individuals. <i>Journal of Investigative Dermatology</i> , <b>2018</b> , 138, 2690-2693	4.3	16
516	Black-White Differences in Housing Type and Sleep Duration as Well as Sleep Difficulties in the United States. <i>International Journal of Environmental Research and Public Health</i> , <b>2018</b> , 15,	4.6	21
515	Next generation sequencing identifies double homozygous mutations in two distinct genes (EXPH5 and COL17A1) in a patient with concomitant simplex and junctional epidermolysis bullosa. <i>Human Mutation</i> , <b>2018</b> , 39, 1349-1354	4.7	19
514	Clinical subtypes and molecular basis of epidermolysis bullosa in Kuwait. <i>International Journal of Dermatology</i> , <b>2018</b> , 57, 1058-1067	1.7	2
513	Efficacy of Human Placental-Derived Stem Cells in Collagen VII Knockout (Recessive Dystrophic Epidermolysis Bullosa) Animal Model. <i>Stem Cells Translational Medicine</i> , <b>2018</b> , 7, 530-542	6.9	6
512	Toll-like Receptor 4 Pathway Polymorphisms Interact with Pollution to Influence Asthma Diagnosis and Severity. <i>Scientific Reports</i> , <b>2018</b> , 8, 12713	4.9	12
511	APOBEC mutation drives early-onset squamous cell carcinomas in recessive dystrophic epidermolysis bullosa. <i>Science Translational Medicine</i> , <b>2018</b> , 10,	17.5	53
510	Reduced Intensity Conditioned Bone Marrow Transplant with Post-Transplant Cyclophosphamide and Donor-Derived Mesenchymal Stromal Cell Infusions for Recessive Dystrophic Epidermolysis Bullosa. <i>Blood</i> , <b>2018</b> , 132, 2165-2165	2.2	1
509	Recessive mutation in tetraspanin CD151 causes Kindler syndrome-like epidermolysis bullosa with multi-systemic manifestations including nephropathy. <i>Matrix Biology</i> , <b>2018</b> , 66, 22-33	11.4	39
508	Robot-assisted radical cystectomy with intracorporeal urinary diversion versus open radical cystectomy (iROC): protocol for a randomised controlled trial with internal feasibility study. <i>BMJ Open</i> , <b>2018</b> , 8, e020500	3	54

507	Genome-wide meta-analysis implicates mediators of hair follicle development and morphogenesis in risk for severe acne. <i>Nature Communications</i> , <b>2018</b> , 9, 5075	17.4	18
506	Cord Blood-Derived Stem Cells Suppress Fibrosis and May Prevent Malignant Progression in Recessive Dystrophic Epidermolysis Bullosa. <i>Stem Cells</i> , <b>2018</b> , 36, 1839-1850	5.8	11
505	Lichen planus and lichenoid dermatoses: Clinical overview and molecular basis. <i>Journal of the American Academy of Dermatology</i> , <b>2018</b> , 79, 789-804	4.5	54
504	Lichen planus and lichenoid dermatoses: Conventional and emerging therapeutic strategies. <i>Journal of the American Academy of Dermatology</i> , <b>2018</b> , 79, 807-818	4.5	19
503	Multidomain Quantitative Recovery Following Radical Cystectomy for Patients Within the Robot-assisted Radical Cystectomy with Intracorporeal Urinary Diversion Versus Open Radical Cystectomy Randomised Controlled Trial: The First 30 Patients. <i>European Urology</i> , <b>2018</b> , 74, 531-534	10.2	21
502	A randomised trial of observational learning from 2D and 3D models in robotically assisted surgery. <i>Surgical Endoscopy and Other Interventional Techniques</i> , <b>2018</b> , 32, 4527-4532	5.2	2
501	Systematised naevus sebaceus resulting from post-zygotic mutation in HRAS. <i>Australasian Journal of Dermatology</i> , <b>2017</b> , 58, 58-60	1.3	3
500	Alopecia, palmoplantar keratoderma, skin fragility and follicular hyperkeratoses due to compound heterozygous mutations in desmoplakin. <i>Australasian Journal of Dermatology</i> , <b>2017</b> , 58, e17-e19	1.3	3
499	Predictive phenotyping of inherited ichthyosis by next-generation DNA sequencing. <i>British Journal of Dermatology</i> , <b>2017</b> , 176, 249-251	4	6
498	Mutations in KLHL24 Add to the Molecular Heterogeneity of Epidermolysis Bullosa Simplex. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, 1378-1380	4.3	30
497	Parastomal hernia following cystectomy and ileal conduit urinary diversion: a systematic review. <i>Hernia: the Journal of Hernias and Abdominal Wall Surgery</i> , <b>2017</b> , 21, 163-175	3.2	28
496	Congenital Anonychia and Uncombable Hair Syndrome: Coinheritance of Homozygous Mutations in RSPO4 and PADI3. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, 1176-1179	4.3	13
495	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 364-370	11	22
494	The Molecular Revolution in Cutaneous Biology: Era of Molecular Diagnostics for Inherited Skin Diseases. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, e83-e86	4.3	7
493	The Gulf Long-Term Follow-Up Study (GuLF STUDY): Biospecimen collection at enrollment. <i>Journal of Toxicology and Environmental Health - Part A: Current Issues</i> , <b>2017</b> , 80, 218-229	3.2	18
492	A new clinical diagnostic matrix for epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2017</b> , 176, 1442-1443		
491	WNT10A mutation causes ectodermal dysplasia by impairing progenitor cell proliferation and KLF4-mediated differentiation. <i>Nature Communications</i> , <b>2017</b> , 8, 15397	17.4	58
490	Amlexanox Enhances Premature Termination Codon Read-Through in COL7A1 and Expression of Full Length Type VII Collagen: Potential Therapy for Recessive Dystrophic Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, 1842-1849	4.3	52

489	PLACK syndrome resulting from a new homozygous insertion mutation in CAST. <i>Journal of Dermatological Science</i> , <b>2017</b> , 88, 256-258	4.3	6
488	Deficient stratum corneum intercellular lipid in a Japanese patient with lamellar ichthyosis with a homozygous deletion mutation in SDR9C7. <i>British Journal of Dermatology</i> , <b>2017</b> , 177, e62-e64	4	15
487	ATR gene mutations in HPV negative oropharyngeal cancer. <i>Oral Oncology</i> , <b>2017</b> , 65, 121-123	4.4	1
486	Surgery: Enhanced recovery after cystectomy: cocktails, culture, or consistency?. <i>Nature Reviews Urology</i> , <b>2017</b> , 14, 648-649	5.5	1
485	Novel and emerging therapies in the treatment of recessive dystrophic epidermolysis bullosa. <i>Intractable and Rare Diseases Research</i> , <b>2017</b> , 6, 6-20	1.4	56
484	Novel homozygous missense mutation in NT5C2 underlying hereditary spastic paraplegia SPG45. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 3109-3113	2.5	11
483	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in a Spectrum of Keratinization Disorders Associated with Thrombocytopenia. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, 2344-2353	4.3	35
482	Tissue and Circulating MicroRNA Co-expression Analysis Shows Potential Involvement of miRNAs in the Pathobiology of Frontal Fibrosing Alopecia. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, 2440-2443	4.3	7
481	Advances in the genetic understanding of hypohidrotic ectodermal dysplasia. <i>Expert Opinion on Orphan Drugs</i> , <b>2017</b> , 5, 967-975	1.1	1
480	Introduction of robot-assisted radical cystectomy within an established enhanced recovery programme. <i>BJU International</i> , <b>2017</b> , 120, 265-272	5.6	13
479	Pityriasis Rubra Pilaris Type V as an Autoinflammatory Disease by CARD14 Mutations. <i>JAMA Dermatology</i> , <b>2017</b> , 153, 66-70	5.1	43
478	Syndromic inherited poikiloderma due to a de novo mutation in FAM111B. <i>British Journal of Dermatology</i> , <b>2017</b> , 176, 534-536	4	9
477	Further evidence for genotype-phenotype disparity in Griscelli syndrome. <i>British Journal of Dermatology</i> , <b>2017</b> , 176, 1086-1089	4	5
476	Questioning the Clinical Utility of Exome Sequencing in Developing Countries. <i>Pediatric Dermatology</i> , <b>2017</b> , 34, e32-e34	1.9	
475	Mental health indicators associated with oil spill response and clean-up: cross-sectional analysis of the GuLF STUDY cohort. <i>Lancet Public Health, The</i> , <b>2017</b> , 2, e560-e567	22.4	27
474	Gene Therapy for Inherited Skin Disorders <b>2017</b> , 1-15		6
473	The effect of observing novice and expert performance on acquisition of surgical skills on a robotic platform. <i>PLoS ONE</i> , <b>2017</b> , 12, e0188233	3.7	9
472	Novel missense mutation in a patient with recessive pretibial epidermolysis bullosa and a mild phenotype. <i>Journal of the European Academy of Dermatology and Venereology</i> , <b>2016</b> , 30, e115-e116	4.6	5

471	Ectodermal dysplasia-skin fragility syndrome resulting from a new atypical homozygous cryptic acceptor splice site mutation in PKP1. <i>Journal of Dermatological Science</i> , <b>2016</b> , 84, 210-212	4.3	2
470	Ichthyosis Prematurity Syndrome: From Fetus to Adulthood. <i>JAMA Dermatology</i> , <b>2016</b> , 152, 1055-8	5.1	6
469	Kindlin-1 Regulates Keratinocyte Electrotaxis. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 2229-2239.	4.3	9
468	Germline NLRP1 Mutations Cause Skin Inflammatory and Cancer Susceptibility Syndromes via Inflammasome Activation. <i>Cell</i> , <b>2016</b> , 167, 187-202.e17	56.2	224
467	Frontal fibrosing alopecia: there is no statistically significant association with leave-on facial skin care products and sunscreens. <i>British Journal of Dermatology</i> , <b>2016</b> , 175, 1407-1408	4	12
466	Management of cutaneous squamous cell carcinoma in patients with epidermolysis bullosa: best clinical practice guidelines. <i>British Journal of Dermatology</i> , <b>2016</b> , 174, 56-67	4	70
465	Intrafamilial phenotypic heterogeneity of epidermolytic ichthyosis associated with a new missense mutation in keratin 10. <i>Clinical and Experimental Dermatology</i> , <b>2016</b> , 41, 290-3	1.8	3
464	Large Intragenic KRT1 Deletion Underlying Atypical Autosomal Dominant Keratinopathic Ichthyosis. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 2095-2098	4.3	3
463	Progress toward Treatment and Cure of Epidermolysis Bullosa: Summary of the DEBRA International Research Symposium EB2015. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 352-358	4.3	48
462	Bi-allelic nonsense mutations in ABHD5 underlie a mild phenotype of Dorfman-Chanarin syndrome. <i>Journal of Dermatological Science</i> , <b>2016</b> , 81, 134-6	4.3	12
461	Venturing into the New Science of Nucleases. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 742-745	4.3	3
460	The p.Glu477Lys Mutation in Keratin 5 Is Strongly Associated with Mortality in Generalized Severe Epidermolysis Bullosa Simplex. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 719-721	4.3	15
459	Lentiviral Engineered Fibroblasts Expressing Codon-Optimized COL7A1 Restore Anchoring Fibrils in RDEB. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 284-92	4.3	34
458	Suppression of TGF $\beta$ and Angiogenesis by Type VII Collagen in Cutaneous SCC. <i>Journal of the National Cancer Institute</i> , <b>2016</b> , 108,	9.7	48
457	Erythrokeratoderma Variabilis Caused by p.Gly45Glu in Connexin 31: Importance of the First Extracellular Loop Glycine Residue for Gap Junction Function. <i>Acta Dermato-Venereologica</i> , <b>2016</b> , 96, 557-9	2.2	3
456	Autosomal dominant diffuse nonepidermolytic palmoplantar keratoderma due to a recurrent mutation in aquaporin-5. <i>British Journal of Dermatology</i> , <b>2016</b> , 174, 430-2	4	7
455	Mutations in EXPH5 underlie a rare subtype of autosomal recessive epidermolysis bullosa simplex. <i>British Journal of Dermatology</i> , <b>2016</b> , 174, 452-3	4	7
454	Cardiomyopathy diagnosed in the eldest child harbouring p.S24X mutation in JUP. <i>British Journal of Dermatology</i> , <b>2016</b> , 175, 644-6	4	8

453	Improved molecular diagnosis of the common recurrent intragenic deletion mutation in IKBKG in a Filipino family with incontinentia pigmenti. <i>Australasian Journal of Dermatology</i> , <b>2016</b> , 57, 150-3	1.3	2
452	Incontinentia pigmenti in a father and daughter. <i>British Journal of Dermatology</i> , <b>2016</b> , 175, 1059-1060	4	9
451	Finasteride is of uncertain utility in treating frontal fibrosing alopecia. <i>Journal of the American Academy of Dermatology</i> , <b>2016</b> , 74, e73-4	4.5	8
450	Enhanced Recovery After Robot-assisted Radical Cystectomy: EAU Robotic Urology Section Scientific Working Group Consensus View. <i>European Urology</i> , <b>2016</b> , 70, 649-660	10.2	90
449	Frontal fibrosing alopecia: reflections and hypotheses on aetiology and pathogenesis. <i>Experimental Dermatology</i> , <b>2016</b> , 25, 847-852	4	57
448	Induced Pluripotent Stem Cell Differentiation and Three-Dimensional Tissue Formation Attenuate Clonal Epigenetic Differences in Trichohyalin. <i>Stem Cells and Development</i> , <b>2016</b> , 25, 1366-75	4.4	8
447	Olmsted syndrome in an Indian male with a new de novo mutation in TRPV3. <i>British Journal of Dermatology</i> , <b>2016</b> , 174, 209-11	4	10
446	Most individuals with either segmental or non-segmental vitiligo display evidence of bilateral cochlear dysfunction. <i>British Journal of Dermatology</i> , <b>2015</b> , 172, 406-11	4	16
445	The three-body problem of therapy with induced pluripotent stem cells. <i>Genome Medicine</i> , <b>2015</b> , 7, 15	14.4	5
444	Whole-exome sequencing diagnosis of two autosomal recessive disorders in one family. <i>British Journal of Dermatology</i> , <b>2015</b> , 172, 1407-11	4	16
443	Familial frontal fibrosing alopecia. <i>Journal of the American Academy of Dermatology</i> , <b>2015</b> , 73, e37	4.5	15
442	Trends in operative caseload and mortality rates after radical cystectomy for bladder cancer in England for 1998-2010. <i>European Urology</i> , <b>2015</b> , 67, 1056-1062	10.2	47
441	Activating CARD14 Mutations Are Associated with Generalized Pustular Psoriasis but Rarely Account for Familial Recurrence in Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , <b>2015</b> , 135, 2964-2970	4.3	58
440	Potential of Systemic Allogeneic Mesenchymal Stromal Cell Therapy for Children with Recessive Dystrophic Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , <b>2015</b> , 135, 2319-2321	4.3	89
439	Improving the understanding of the link between cognition and functional capacity in schizophrenia and bipolar disorder. <i>Schizophrenia Research</i> , <b>2015</b> , 169, 121-127	3.6	11
438	The cytolinker plectin regulates nuclear mechanotransduction in keratinocytes. <i>Journal of Cell Science</i> , <b>2015</b> , 128, 4475-86	5.3	30
437	Novel indel mutation of STS underlies a new phenotype of self-healing recessive X-linked ichthyosis. <i>Journal of Dermatological Science</i> , <b>2015</b> , 79, 317-9	4.3	5
436	Progressive hyperpigmentation in a Taiwanese child due to an inborn error of vitamin B12 metabolism (cblJ). <i>British Journal of Dermatology</i> , <b>2015</b> , 172, 1111-5	4	17

435	Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. <i>British Journal of Dermatology</i> , <b>2015</b> , 172, 94-100	4	54
434	EGFR mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2015</b> , 3, 452-8	2.3	8
433	Over the horizon - future innovations in global urology. <i>BJU International</i> , <b>2015</b> , 116, 318-20	5.6	3
432	Urodynamics For Prostate Surgery Trial; Randomised Evaluation of Assessment Methods (UPSTREAM) for diagnosis and management of bladder outlet obstruction in men: study protocol for a randomised controlled trial. <i>Trials</i> , <b>2015</b> , 16, 567	2.8	30
431	Founder mutation in dystonin-e underlying autosomal recessive epidermolysis bullosa simplex in Kuwait. <i>British Journal of Dermatology</i> , <b>2015</b> , 172, 527-31	4	20
430	Global surgery - how much of the burden is urological?. <i>BJU International</i> , <b>2015</b> , 116, 314-6	5.6	9
429	Current challenges to urological training in sub-Saharan Africa. <i>BJU International</i> , <b>2015</b> , 116, 316-8	5.6	4
428	Ichthyosis follicularis, atrichia, and photophobia syndrome associated with a new mutation in MBTPS2. <i>Clinical and Experimental Dermatology</i> , <b>2015</b> , 40, 529-32	1.8	12
427	Familial progressive hyper- and hypopigmentation and malignancy in two families with new mutations in KITLG. <i>Clinical and Experimental Dermatology</i> , <b>2015</b> , 40, 860-4	1.8	11
426	Genome-wide association study of schizophrenia in Ashkenazi Jews. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2015</b> , 168, 649-59	3.5	128
425	Novel TGM5 mutations in acral peeling skin syndrome. <i>Experimental Dermatology</i> , <b>2015</b> , 24, 285-9	4	10
424	Is adermatoglyphia an additional feature of Kindler Syndrome?. <i>Anais Brasileiros De Dermatologia</i> , <b>2015</b> , 90, 592-3	1.6	3
423	Lipoid proteinosis. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , <b>2015</b> , 132, 317-23		14
422	Recently Identified Forms of Epidermolysis Bullosa. <i>Annals of Dermatology</i> , <b>2015</b> , 27, 658-66	0.4	30
421	Familial primary localized cutaneous amyloidosis results from either dominant or recessive mutations in OSMR. <i>Acta Dermato-Venereologica</i> , <b>2015</b> , 95, 1005-7	2.2	8
420	Novel autosomal dominant mutation in loricrin presenting as prominent ichthyosis. <i>British Journal of Dermatology</i> , <b>2015</b> , 173, 1291-4	4	12
419	Reduced Toxicity Conditioning and Allogeneic Hematopoietic Progenitor Cell Transplantation for Recessive Dystrophic Epidermolysis Bullosa. <i>Journal of Pediatrics</i> , <b>2015</b> , 167, 765-9.e1	3.6	21
418	Diagnosis by numbers: defining skin disease pathogenesis through collated gene signatures. <i>Journal of Investigative Dermatology</i> , <b>2015</b> , 135, 17-19	4.3	2

417	Multicenter assessment of neoadjuvant chemotherapy for muscle-invasive bladder cancer. <i>European Urology</i> , <b>2015</b> , 67, 241-9	10.2	178
416	Anesthesia for major urologic surgery. <i>Anesthesiology Clinics</i> , <b>2015</b> , 33, 165-72	2.3	11
415	Transplanted bone marrow-derived circulating PDGFR $\beta$ cells restore type VII collagen in recessive dystrophic epidermolysis bullosa mouse skin graft. <i>Journal of Immunology</i> , <b>2015</b> , 194, 1996-2003	5.3	52
414	Infection and inflammation in schizophrenia and bipolar disorder: a genome wide study for interactions with genetic variation. <i>PLoS ONE</i> , <b>2015</b> , 10, e0116696	3.7	73
413	Lysyl Hydroxylase 3 Localizes to Epidermal Basement Membrane and Is Reduced in Patients with Recessive Dystrophic Epidermolysis Bullosa. <i>PLoS ONE</i> , <b>2015</b> , 10, e0137639	3.7	18
412	Ectodermal Dysplasia Skin Fragility Syndrome <b>2015</b> , 307-312		
411	Desmosomal Proteins and Their Role in Epidermolysis Bullosa <b>2015</b> , 49-54		1
410	Kindler Syndrome <b>2015</b> , 433-439		1
409	Targeted prostate cancer screening in BRCA1 and BRCA2 mutation carriers: results from the initial screening round of the IMPACT study. <i>European Urology</i> , <b>2014</b> , 66, 489-99	10.2	156
408	Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. <i>Journal of the American Academy of Dermatology</i> , <b>2014</b> , 70, 1103-26	4.5	596
407	Under-recognition of acral peeling skin syndrome: 59 new cases with 15 novel mutations. <i>British Journal of Dermatology</i> , <b>2014</b> , 171, 1206-10	4	21
406	Patient-specific naturally gene-reverted induced pluripotent stem cells in recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>2014</b> , 134, 1246-1254	4.3	63
405	Inherited blistering skin diseases: underlying molecular mechanisms and emerging therapies. <i>Annals of Medicine</i> , <b>2014</b> , 46, 49-61	1.5	9
404	Treatment of hereditary epidermolysis bullosa: updates and future prospects. <i>American Journal of Clinical Dermatology</i> , <b>2014</b> , 15, 1-6	7.1	39
403	Naevus sebaceus: a mosaic RASopathy. <i>Clinical and Experimental Dermatology</i> , <b>2014</b> , 39, 1-6	1.8	36
402	New intragenic and promoter region deletion mutations in FERMT1 underscore genetic homogeneity in Kindler syndrome. <i>Clinical and Experimental Dermatology</i> , <b>2014</b> , 39, 361-7	1.8	11
401	Olmsted syndrome in an Iranian boy with a new de novo mutation in TRPV3. <i>Clinical and Experimental Dermatology</i> , <b>2014</b> , 39, 492-5	1.8	13
400	Enhanced recovery programmes for patients undergoing radical cystectomy. <i>Nature Reviews Urology</i> , <b>2014</b> , 11, 437-44	5.5	18

399	BPAG1-e restricts keratinocyte migration through control of adhesion stability. <i>Journal of Investigative Dermatology</i> , <b>2014</b> , 134, 773-782	4.3	25
398	Epithelial inflammation resulting from an inherited loss-of-function mutation in EGFR. <i>Journal of Investigative Dermatology</i> , <b>2014</b> , 134, 2570-2578	4.3	51
397	3D In vitro model of a functional epidermal permeability barrier from human embryonic stem cells and induced pluripotent stem cells. <i>Stem Cell Reports</i> , <b>2014</b> , 2, 675-89	8	86
396	Cell therapy in dermatology. <i>Cold Spring Harbor Perspectives in Medicine</i> , <b>2014</b> , 4,	5.4	20
395	Preconditioning of mesenchymal stem cells for improved transplantation efficacy in recessive dystrophic epidermolysis bullosa. <i>Stem Cell Research and Therapy</i> , <b>2014</b> , 5, 121	8.3	32
394	Type VII collagen regulates expression of OATP1B3, promotes front-to-rear polarity and increases structural organisation in 3D spheroid cultures of RDEB tumour keratinocytes. <i>Journal of Cell Science</i> , <b>2014</b> , 127, 740-51	5.3	14
393	Functional variants in DPYSL2 sequence increase risk of schizophrenia and suggest a link to mTOR signaling. <i>G3: Genes, Genomes, Genetics</i> , <b>2014</b> , 5, 61-72	3.2	29
392	Lipoid proteinosis: rare case confirmed by ECM1 mutation detection. <i>International Journal of Pediatric Otorhinolaryngology</i> , <b>2014</b> , 78, 2314-5	1.7	3
391	Clinical features and WNT10A mutations in seven unrelated cases of Schöpf-Schulz-Passarge syndrome. <i>British Journal of Dermatology</i> , <b>2014</b> , 171, 1211-4	4	20
390	Wharton's jelly mesenchymal stromal/stem cells derived under chemically defined animal product-free low oxygen conditions are rich in MSCA-1(+) subpopulation. <i>Regenerative Medicine</i> , <b>2014</b> , 9, 723-32	2.5	12
389	The missense mutation p.R1303Q in type XVII collagen underlies junctional epidermolysis bullosa resembling Kindler syndrome. <i>Journal of Investigative Dermatology</i> , <b>2014</b> , 134, 845-849	4.3	16
388	Keratin 9 is required for the structural integrity and terminal differentiation of the palmoplantar epidermis. <i>Journal of Investigative Dermatology</i> , <b>2014</b> , 134, 754-763	4.3	55
387	Augmentation of cutaneous wound healing by pharmacologic mobilization of endogenous bone marrow stem cells. <i>Journal of Investigative Dermatology</i> , <b>2014</b> , 134, 2312-2314	4.3	5
386	Mutations in GRHL2 result in an autosomal-recessive ectodermal Dysplasia syndrome. <i>American Journal of Human Genetics</i> , <b>2014</b> , 95, 308-14	11	37
385	High levels of type VII collagen expression in recessive dystrophic epidermolysis bullosa cutaneous squamous cell carcinoma keratinocytes increases PI3K and MAPK signalling, cell migration and invasion. <i>British Journal of Dermatology</i> , <b>2014</b> , 170, 1256-65	4	14
384	Limited ectrodactyly, ectodermal dysplasia and cleft lip-palate syndrome with a p63 mutation, associated with linear and whorled naevoid hypermelanosis. <i>Clinical and Experimental Dermatology</i> , <b>2014</b> , 39, 266-8	1.8	1
383	A Scandinavian case of skin fragility, alopecia and cardiomyopathy caused by DSP mutations. <i>Clinical and Experimental Dermatology</i> , <b>2014</b> , 39, 30-4	1.8	8
382	Evolution of the Southampton Enhanced Recovery Programme for radical cystectomy and the aggregation of marginal gains. <i>BJU International</i> , <b>2014</b> , 114, 375-83	5.6	38

381	On Medawar's 'Actively acquired tolerance of foreign cells'. <i>Experimental Dermatology</i> , <b>2014</b> , 23, 97-8	4	1
380	Familial carotenaemia and carotenoderma. <i>Clinical and Experimental Dermatology</i> , <b>2014</b> , 39, 771-2	1.8	2
379	Mutations in EXPH5 result in autosomal recessive inherited skin fragility. <i>British Journal of Dermatology</i> , <b>2014</b> , 170, 196-9	4	9
378	Implementation of the Exeter enhanced recovery programme for patients undergoing radical cystectomy. <i>BJU International</i> , <b>2014</b> , 113, 719-25	5.6	41
377	Association of obesity and treated hypertension and diabetes with cognitive ability in bipolar disorder and schizophrenia. <i>Bipolar Disorders</i> , <b>2014</b> , 16, 422-31	3.8	53
376	Somatic forward (nonrevertant) mosaicism in recessive dystrophic epidermolysis bullosa. <i>JAMA Dermatology</i> , <b>2014</b> , 150, 1025-7	5.1	6
375	Next generation diagnostics of heritable connective tissue disorders. <i>Matrix Biology</i> , <b>2014</b> , 33, 35-40	11.4	9
374	Reciprocal duplication of the Williams-Beuren syndrome deletion on chromosome 7q11.23 is associated with schizophrenia. <i>Biological Psychiatry</i> , <b>2014</b> , 75, 371-7	7.9	54
373	Kindlin-1 controls Wnt and TGF- $\beta$ availability to regulate cutaneous stem cell proliferation. <i>Nature Medicine</i> , <b>2014</b> , 20, 350-9	50.5	101
372	Gene therapies for inherited skin disorders. <i>Seminars in Cutaneous Medicine and Surgery</i> , <b>2014</b> , 33, 83-90	1.4	15
371	Blasenkarzinom bei Frauen. <i>Praxis</i> , <b>2014</b> , 103, 1149-1152	0.1	
370	Late diagnosis of ectodermal dysplasia syndrome. <i>Australasian Journal of Dermatology</i> , <b>2013</b> , 54, 46-8	1.3	5
369	Acral peeling skin syndrome resulting from a homozygous nonsense mutation in the CSTA gene encoding cystatin A. <i>Pediatric Dermatology</i> , <b>2013</b> , 30, e87-8	1.9	30
368	Structure and function of skin, hair and nails. <i>Medicine</i> , <b>2013</b> , 41, 317-320	0.6	40
367	Impact of next generation sequencing on diagnostics in a genetic skin disease clinic. <i>Experimental Dermatology</i> , <b>2013</b> , 22, 825-31	4	41
366	Serum levels of high mobility group box 1 correlate with disease severity in recessive dystrophic epidermolysis bullosa. <i>Experimental Dermatology</i> , <b>2013</b> , 22, 433-5	4	22
365	Fibroblast cell therapy enhances initial healing in recessive dystrophic epidermolysis bullosa wounds: results of a randomized, vehicle-controlled trial. <i>British Journal of Dermatology</i> , <b>2013</b> , 169, 1025-33	4.33	92
364	Multiple variants aggregate in the neuregulin signaling pathway in a subset of schizophrenia patients. <i>Translational Psychiatry</i> , <b>2013</b> , 3, e264	8.6	31

363	Phase I study protocol for ex vivo lentiviral gene therapy for the inherited skin disease, Netherton syndrome. <i>Human Gene Therapy Clinical Development</i> , <b>2013</b> , 24, 182-90	3.2	30
362	Epidermolysis bullosa pruriginosa: a case with prominent histopathologic inflammation. <i>JAMA Dermatology</i> , <b>2013</b> , 149, 727-31	5.1	16
361	Increasing research capacity and capability - how can national infrastructure help?. <i>BJU International</i> , <b>2013</b> , 111, 370-1	5.6	1
360	Late-onset pretibial recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , <b>2013</b> , 38, 630-2	1.8	6
359	A new homozygous nonsense mutation in LAMA3A underlying laryngo-onycho-cutaneous syndrome. <i>British Journal of Dermatology</i> , <b>2013</b> , 169, 1353-6	4	15
358	Hyaline fibromatosis syndrome resulting from a new homozygous missense mutation, p.Gly116Val, in ANTXR2. <i>Journal of Dermatology</i> , <b>2013</b> , 40, 677-8	1.6	4
357	Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. <i>Nature Genetics</i> , <b>2013</b> , 45, 1244-1248	36.3	217
356	G28 A Novel Missense Mutation in Keratin 1 Underlying Clinically Mild Epidermolytic Ichthyosis Mimicking Epidermolysis Bullosa Simplex Superficialis. <i>Archives of Disease in Childhood</i> , <b>2013</b> , 98, A17-A18 <sup>2</sup>		
355	Sporadic Kindler syndrome with a novel mutation. <i>Anais Brasileiros De Dermatologia</i> , <b>2013</b> , 88, 212-5	1.6	7
354	Revertant mosaicism in the skin. <i>Giornale Italiano Di Dermatologia E Venereologia</i> , <b>2013</b> , 148, 73-82	0.8	3
353	Determinants of occupational and residential functioning in bipolar disorder. <i>Journal of Affective Disorders</i> , <b>2012</b> , 136, 812-8	6.6	20
352	Case of Kindler syndrome resulting from mutation in the FERMT1 gene. <i>Journal of Dermatology</i> , <b>2012</b> , 39, 1057-8	1.6	6
351	Skin differences based on age and chronicity of ultraviolet exposure: results from a gene expression profiling study. <i>British Journal of Dermatology</i> , <b>2012</b> , 166 Suppl 2, 9-15	4	21
350	Twenty top tips to triumph in dermatology. <i>British Journal of Dermatology</i> , <b>2012</b> , 167, 445-6	4	
349	Mutations in AEC syndrome skin reveal a role for p63 in basement membrane adhesion, skin barrier integrity and hair follicle biology. <i>British Journal of Dermatology</i> , <b>2012</b> , 167, 134-44	4	22
348	Recurrent heterozygous missense mutation, p.Gly573Ser, in the TRPV3 gene in an Indian boy with sporadic Olmsted syndrome. <i>British Journal of Dermatology</i> , <b>2012</b> , 167, 440-2	4	55
347	MBTPS2 mutation in a British pedigree with keratosis follicularis spinulosa decalvans. <i>Clinical and Experimental Dermatology</i> , <b>2012</b> , 37, 631-4	1.8	12
346	Revertant mosaicism in Kindler syndrome. <i>Journal of Investigative Dermatology</i> , <b>2012</b> , 132, 730-2	4.3	25

345	Limbal stem cell deficiency and ocular phenotype in ectrodactyly-ectodermal dysplasia-clefting syndrome caused by p63 mutations. <i>Ophthalmology</i> , <b>2012</b> , 119, 74-83	7.3	63
344	Acral peeling skin syndrome: a clinically and genetically heterogeneous disorder. <i>Pediatric Dermatology</i> , <b>2012</b> , 29, 258-63	1.9	11
343	Fibroblast-derived dermal matrix drives development of aggressive cutaneous squamous cell carcinoma in patients with recessive dystrophic epidermolysis bullosa. <i>Cancer Research</i> , <b>2012</b> , 72, 3522-34	10.1	83
342	Perioperative fluid management: Consensus statement from the enhanced recovery partnership. <i>Perioperative Medicine (London, England)</i> , <b>2012</b> , 1, 2	2.8	75
341	Novel and recurrent COL7A1 mutations in Chilean patients with dystrophic epidermolysis bullosa. <i>Journal of Dermatological Science</i> , <b>2012</b> , 65, 149-52	4.3	14
340	Germline Mutation in EXPH5 Implicates the Rab27B Effector Protein Slac2-b in Inherited Skin Fragility. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 1115-21	11	48
339	Bone marrow transplantation in epidermolysis bullosa. <i>Immunotherapy</i> , <b>2012</b> , 4, 1859-67	3.8	5
338	Germline mutation in ATR in autosomal- dominant oropharyngeal cancer syndrome. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 511-7	11	49
337	Ectodermal dysplasia-skin fragility syndrome due to a new homozygous internal deletion mutation in the PKP1 gene. <i>Australasian Journal of Dermatology</i> , <b>2012</b> , 53, 61-5	1.3	20
336	Desmosomal genodermatoses. <i>British Journal of Dermatology</i> , <b>2012</b> , 166, 36-45	4	59
335	Infantile systemic hyalinosis associated with a putative splice-site mutation in the ANTXR2 gene. <i>Clinical and Experimental Dermatology</i> , <b>2012</b> , 37, 635-8	1.8	9
334	Autosomal recessive epidermolysis bullosa simplex due to loss of BPAG1-e expression. <i>Journal of Investigative Dermatology</i> , <b>2012</b> , 132, 742-4	4.3	39
333	Novel molecular therapies for heritable skin disorders. <i>Journal of Investigative Dermatology</i> , <b>2012</b> , 132, 820-8	4.3	49
332	Next-generation diagnostics for genodermatoses. <i>Journal of Investigative Dermatology</i> , <b>2012</b> , 132, E27-8	4.3	5
331	Profilaggrin, dry skin, and atopic dermatitis risk: size matters. <i>Journal of Investigative Dermatology</i> , <b>2012</b> , 132, 10-1	4.3	12
330	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. <i>Nature Genetics</i> , <b>2012</b> , 44, 1272-6	36.3	56
329	Impairment in functional capacity as an endophenotype candidate in severe mental illness. <i>Schizophrenia Bulletin</i> , <b>2012</b> , 38, 1318-26	1.3	20
328	The structure and function of skin <b>2012</b> , 1-31		1

327	Specialized techniques in dermatopathology <b>2012</b> , 32-45		1
326	Allogeneic Hematopoietic Cell Transplantation for Severe Epidermolysis Bullosa: Impact of Conditioning Regimen and Stem Cell Source On Outcomes. <i>Blood</i> , <b>2012</b> , 120, 965-965	2.2	
325	Early Re-Resection for T1 Transitional Cell Carcinoma of the Bladder—A Study of Current Practice in the South West of England. <i>British Journal of Medical and Surgical Urology</i> , <b>2011</b> , 4, 18-23		1
324	Sensitivity and specificity of the UCSD Performance-based Skills Assessment (UPSA-B) for identifying functional milestones in schizophrenia. <i>Schizophrenia Research</i> , <b>2011</b> , 132, 165-70	3.6	68
323	Revertant mosaicism in skin: natural gene therapy. <i>Trends in Molecular Medicine</i> , <b>2011</b> , 17, 140-8	11.5	90
322	Schöpf-Schulz-Passarge syndrome resulting from a homozygous nonsense mutation, p.Cys107X, in WNT10A. <i>Australasian Journal of Dermatology</i> , <b>2011</b> , 52, 224-6	1.3	18
321	Extracellular matrix protein 1 autoantibodies in male genital lichen sclerosus. <i>British Journal of Dermatology</i> , <b>2011</b> , 165, 218-9	4	36
320	Integrative mRNA profiling comparing cultured primary cells with clinical samples reveals PLK1 and C20orf20 as therapeutic targets in cutaneous squamous cell carcinoma. <i>Oncogene</i> , <b>2011</b> , 30, 4666-77	9.2	54
319	Linkage analysis of plasma dopamine β-hydroxylase activity in families of patients with schizophrenia. <i>Human Genetics</i> , <b>2011</b> , 130, 635-43	6.3	39
318	Molecular and neurological characterizations of three Saudi families with lipoid proteinosis. <i>BMC Medical Genetics</i> , <b>2011</b> , 12, 31	2.1	15
317	Linkage and association on 8p21.2-p21.1 in schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2011</b> , 156, 188-97	3.5	23
316	The international dystrophic epidermolysis bullosa patient registry: an online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. <i>Human Mutation</i> , <b>2011</b> , 32, 1100-17	4.7	63
315	The inversa type of recessive dystrophic epidermolysis bullosa is caused by specific arginine and glycine substitutions in type VII collagen. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 160-7	5.8	21
314	Epidermolysis bullosa pruriginosa masquerading as psychogenic pruritus. <i>Archives of Dermatology</i> , <b>2011</b> , 147, 956-60		20
313	HB-EGF induces COL7A1 expression in keratinocytes and fibroblasts: possible mechanism underlying allogeneic fibroblast therapy in recessive dystrophic epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , <b>2011</b> , 131, 1771-4	4.3	54
312	PDGFRα-positive cells in bone marrow are mobilized by high mobility group box 1 (HMGB1) to regenerate injured epithelia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2011</b> , 108, 6609-14	11.5	178
311	Novel and recurrent FERMT1 gene mutations in Kindler syndrome. <i>Acta Dermato-Venereologica</i> , <b>2011</b> , 91, 267-70	2.2	20
310	Identical glycine substitution mutations in type VII collagen may underlie both dominant and recessive forms of dystrophic epidermolysis bullosa. <i>Acta Dermato-Venereologica</i> , <b>2011</b> , 91, 262-6	2.2	28

309	Next-generation diagnostics for inherited skin disorders. <i>Journal of Investigative Dermatology</i> , <b>2011</b> , 131, 1971-3	4.3	18
308	Intra-familial variability of ectodermal defects associated with WNT10A mutations. <i>Acta Dermato-Venereologica</i> , <b>2011</b> , 91, 346-7	2.2	12
307	The molecular skin pathology of familial primary localized cutaneous amyloidosis. <i>Experimental Dermatology</i> , <b>2010</b> , 19, 416-23	4	28
306	Blistering skin diseases: a bridge between dermatopathology and molecular biology. <i>Histopathology</i> , <b>2010</b> , 56, 91-9	7.3	8
305	Molecular basis of EEC (ectrodactyly, ectodermal dysplasia, clefting) syndrome: five new mutations in the DNA-binding domain of the TP63 gene and genotype-phenotype correlation. <i>British Journal of Dermatology</i> , <b>2010</b> , 162, 201-7	4	31
304	Development and successful clinical application of preimplantation genetic haplotyping for Herlitz junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2010</b> , 162, 1330-6	4	15
303	Spectrum of mutations in the ANTXR2 (CMG2) gene in infantile systemic hyalinosis and juvenile hyaline fibromatosis. <i>British Journal of Dermatology</i> , <b>2010</b> , 163, 213-5	4	35
302	Rapp-Hodgkin and Hay-Wells ectodermal dysplasia syndromes represent a variable spectrum of the same genetic disorder. <i>British Journal of Dermatology</i> , <b>2010</b> , 163, 624-9	4	29
301	Matrix metalloproteinase-7 activates heparin-binding epidermal growth factor-like growth factor in cutaneous squamous cell carcinoma. <i>British Journal of Dermatology</i> , <b>2010</b> , 163, 726-35	4	54
300	Stem cell therapies for recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2010</b> , 163, 1149-56	4	19
299	Bullous pemphigoid in a patient with suspected non-Herlitz junctional epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , <b>2010</b> , 35, 881-4	1.8	2
298	Novel IL31RA gene mutation and ancestral OSMR mutant allele in familial primary cutaneous amyloidosis. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 26-32	5.3	39
297	New insights into hereditary angio-oedema: Molecular diagnosis and therapy. <i>Australasian Journal of Dermatology</i> , <b>2010</b> , 51, 157-62	1.3	7
296	Categorizing immunofluorescence mapping in epidermolysis bullosa with pyloric atresia: Use as a broad prognostic indicator. <i>Australasian Journal of Dermatology</i> , <b>2010</b> , 51, 212-4	1.3	4
295	Relationship of the Brief UCSD Performance-based Skills Assessment (UPSA-B) to multiple indicators of functioning in people with schizophrenia and bipolar disorder. <i>Bipolar Disorders</i> , <b>2010</b> , 12, 45-55	3.8	95
294	Social competence and observer-rated social functioning in bipolar disorder. <i>Bipolar Disorders</i> , <b>2010</b> , 12, 843-50	3.8	37
293	Prediction of real-world functional disability in chronic mental disorders: a comparison of schizophrenia and bipolar disorder. <i>American Journal of Psychiatry</i> , <b>2010</b> , 167, 1116-24	11.9	336
292	Bone marrow transplantation for recessive dystrophic epidermolysis bullosa. <i>New England Journal of Medicine</i> , <b>2010</b> , 363, 629-39	59.2	262

291	Revertant mosaicism in recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>2010</b> , 130, 1937-40	4.3	47
290	Common IL-31 gene haplotype associated with non-atopic eczema is not implicated in epidermolysis bullosa pruriginosa. <i>Acta Dermato-Venereologica</i> , <b>2010</b> , 90, 631-2	2.2	8
289	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. <i>Journal of Investigative Dermatology</i> , <b>2010</b> , 130, 1551-7	4.3	101
288	No evidence that human papillomavirus is responsible for the aggressive nature of recessive dystrophic epidermolysis bullosa-associated squamous cell carcinoma. <i>Journal of Investigative Dermatology</i> , <b>2010</b> , 130, 2853-5	4.3	6
287	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> , <b>2010</b> , 130, 1543-50	4.3	40
286	Progress in epidermolysis bullosa research: toward treatment and cure. <i>Journal of Investigative Dermatology</i> , <b>2010</b> , 130, 1778-84	4.3	57
285	Ectodermal dysplasia-skin fragility syndrome. <i>Dermatologic Clinics</i> , <b>2010</b> , 28, 125-9	4.2	55
284	Kindler syndrome. <i>Dermatologic Clinics</i> , <b>2010</b> , 28, 119-24	4.2	56
283	Schöpf-Schulz-Passarge syndrome resulting from a homozygous nonsense mutation in WNT10A. <i>Journal of Dermatological Science</i> , <b>2010</b> , 58, 220-2	4.3	29
282	Lethal acantholytic epidermolysis bullosa. <i>Dermatologic Clinics</i> , <b>2010</b> , 28, 131-5	4.2	25
281	The role of kindlins in cell biology and relevance to human disease. <i>International Journal of Biochemistry and Cell Biology</i> , <b>2010</b> , 42, 595-603	5.6	65
280	Congenital muscular dystrophy, myasthenic symptoms and epidermolysis bullosa simplex (EBS) associated with mutations in the PLEC1 gene encoding plectin. <i>Neuromuscular Disorders</i> , <b>2010</b> , 20, 709-119	4.9	35
279	Prenatal diagnosis of epidermolysis bullosa. <i>Dermatologic Clinics</i> , <b>2010</b> , 28, 231-7, viii	4.2	22
278	Anatomy and Organization of Human Skin <b>2010</b> , 1-53		30
277	Replication of an association of a common variant in the Reelin gene (RELN) with schizophrenia in Ashkenazi Jewish women. <i>Psychiatric Genetics</i> , <b>2010</b> , 20, 184-6	2.9	37
276	Microdeletions of 3q29 confer high risk for schizophrenia. <i>American Journal of Human Genetics</i> , <b>2010</b> , 87, 229-36	11	173
275	Detection of SNP-SNP interactions in trios of parents with schizophrenic children. <i>Genetic Epidemiology</i> , <b>2010</b> , 34, 396-406	2.6	18
274	Familiality of novel factorial dimensions of schizophrenia. <i>Archives of General Psychiatry</i> , <b>2009</b> , 66, 591-600		61

273	New glycine substitution mutations in type VII collagen underlying epidermolysis bullosa pruriginosa but the phenotype is not explained by a common polymorphism in the matrix metalloproteinase-1 gene promoter. <i>Acta Dermato-Venereologica</i> , <b>2009</b> , 89, 6-11	2.2	29
272	Lipoid proteinosis: identification of two novel mutations in the human ECM-1 gene and lack of genotype-phenotype correlation. <i>Acta Dermato-Venereologica</i> , <b>2009</b> , 89, 528-9	2.2	17
271	Increased invasive behaviour in cutaneous squamous cell carcinoma with loss of basement-membrane type VII collagen. <i>Journal of Cell Science</i> , <b>2009</b> , 122, 1788-99	5.3	81
270	Structure and function of skin, hair and nails. <i>Medicine</i> , <b>2009</b> , 37, 223-226	0.6	35
269	Novel truncating mutations in PKP1 and DSP cause similar skin phenotypes in two Brazilian families. <i>British Journal of Dermatology</i> , <b>2009</b> , 160, 692-7	4	38
268	Acantholytic ectodermal dysplasia: clinicopathological study of a new desmosomal disorder. <i>British Journal of Dermatology</i> , <b>2009</b> , 160, 868-74	4	13
267	Kindler syndrome: a focal adhesion genodermatosis. <i>British Journal of Dermatology</i> , <b>2009</b> , 160, 233-42	4	81
266	Autosomal dominant junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2009</b> , 160, 1094-7	4	28
265	PORCN gene mutations and the protean nature of focal dermal hypoplasia. <i>British Journal of Dermatology</i> , <b>2009</b> , 160, 1103-9	4	32
264	New insight into mechanisms of pruritus from molecular studies on familial primary localized cutaneous amyloidosis. <i>British Journal of Dermatology</i> , <b>2009</b> , 161, 1217-24	4	50
263	Epidermolysis bullosa pruriginosa in association with lichen planopilaris. <i>Clinical and Experimental Dermatology</i> , <b>2009</b> , 34, e825-8	1.8	10
262	Fine mapping on chromosome 10q22-q23 implicates Neuregulin 3 in schizophrenia. <i>American Journal of Human Genetics</i> , <b>2009</b> , 84, 21-34	11	78
261	A novel OSMR mutation in familial primary localized cutaneous amyloidosis in a Japanese family. <i>Journal of Dermatological Science</i> , <b>2009</b> , 55, 64-5	4.3	7
260	Recurrent European missense mutation in the F12 gene in a British family with type III hereditary angioedema. <i>Journal of Dermatological Science</i> , <b>2009</b> , 56, 62-4	4.3	21
259	Loss-of-function FERMT1 mutations in kindler syndrome implicate a role for fermitin family homolog-1 in integrin activation. <i>American Journal of Pathology</i> , <b>2009</b> , 175, 1431-41	5.8	30
258	Molecular Basis of Skin Disease <b>2009</b> , 519-550		
257	The Role of Filaggrin in Skin Diseases. <i>Basic and Clinical Dermatology</i> , <b>2009</b> , 57-68		
256	Colocalization of kindlin-1, kindlin-2, and migfilin at keratinocyte focal adhesion and relevance to the pathophysiology of Kindler syndrome. <i>Journal of Investigative Dermatology</i> , <b>2008</b> , 128, 2156-65	4.3	69

255	Potential of fibroblast cell therapy for recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>2008</b> , 128, 2179-89	4.3	176
254	KRT14 haploinsufficiency results in increased susceptibility of keratinocytes to TNF-alpha-induced apoptosis and causes Naegeli-Franceschetti-Jadassohn syndrome. <i>Journal of Investigative Dermatology</i> , <b>2008</b> , 128, 1517-24	4.3	42
253	Filaggrin and the great epidermal barrier grief. <i>Australasian Journal of Dermatology</i> , <b>2008</b> , 49, 67-73; quiz 73-4	1.3	28
252	Introduction of an enhanced recovery protocol for radical cystectomy. <i>BJU International</i> , <b>2008</b> , 101, 698-701	3.01	148
251	Transformation-specific matrix metalloproteinases (MMP)-7 and MMP-13 are expressed by tumour cells in epidermolysis bullosa-associated squamous cell carcinomas. <i>British Journal of Dermatology</i> , <b>2008</b> , 158, 778-85	4	46
250	Focal dermal hypoplasia resulting from a new nonsense mutation, p.E300X, in the PORCN gene. <i>Journal of Dermatological Science</i> , <b>2008</b> , 49, 39-42	4.3	22
249	The filaggrin story: novel insights into skin-barrier function and disease. <i>Trends in Molecular Medicine</i> , <b>2008</b> , 14, 20-7	11.5	174
248	Oncostatin M receptor-beta mutations underlie familial primary localized cutaneous amyloidosis. <i>American Journal of Human Genetics</i> , <b>2008</b> , 82, 73-80	11	99
247	The classification of inherited epidermolysis bullosa (EB): Report of the Third International Consensus Meeting on Diagnosis and Classification of EB. <i>Journal of the American Academy of Dermatology</i> , <b>2008</b> , 58, 931-50	4.5	690
246	Molecular genetics as a diagnostic and prognostic aid in the assessment of neonates with red, scaly genodermatoses: work still in progress. <i>Archives of Dermatology</i> , <b>2008</b> , 144, 387-8		1
245	Desquamative enteropathy and pyloric atresia without skin disease caused by a novel intracellular beta4 integrin mutation. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2008</b> , 47, 585-91	2.8	18
244	A novel translation re-initiation mechanism for the p63 gene revealed by amino-terminal truncating mutations in Rapp-Hodgkin/Hay-Wells-like syndromes. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 1968-77	5.6	43
243	Avoiding verification bias in screening test evaluation in resource poor settings: a case study from Zimbabwe. <i>Clinical Trials</i> , <b>2008</b> , 5, 496-503	2.2	13
242	A heterozygous frameshift mutation in the V1 domain of keratin 5 in a family with Dowling-Degos disease. <i>Journal of Investigative Dermatology</i> , <b>2007</b> , 127, 298-300	4.3	39
241	The South African "bathing suit ichthyosis" is a form of lamellar ichthyosis caused by a homozygous missense mutation, p.R315L, in transglutaminase 1. <i>Journal of Investigative Dermatology</i> , <b>2007</b> , 127, 490-3	4.3	23
240	Genetic diseases of junctions. <i>Journal of Investigative Dermatology</i> , <b>2007</b> , 127, 2713-25	4.3	113
239	Five new homozygous mutations in the KIND1 gene in Kindler syndrome. <i>Journal of Investigative Dermatology</i> , <b>2007</b> , 127, 2268-70	4.3	24
238	Patients with recessive dystrophic epidermolysis bullosa develop squamous-cell carcinoma regardless of type VII collagen expression. <i>Journal of Investigative Dermatology</i> , <b>2007</b> , 127, 2438-44	4.3	40

237	The role of fibroblasts in tissue engineering and regeneration. <i>British Journal of Dermatology</i> , <b>2007</b> , 156, 1149-55	4	218
236	New KRT10 gene mutation underlying the annular variant of bullous congenital ichthyosiform erythroderma with clinical worsening during pregnancy. <i>British Journal of Dermatology</i> , <b>2007</b> , 157, 602-4 <sup>4</sup>		12
235	Recurrent p.N767S mutation in the ATP2A2 gene in a Japanese family with haemorrhagic Darier disease clinically mimicking epidermolysis bullosa simplex with mottled pigmentation. <i>British Journal of Dermatology</i> , <b>2007</b> , 157, 605-8	4	12
234	Functional redundancy of extracellular matrix protein 1 in epidermal differentiation. <i>British Journal of Dermatology</i> , <b>2007</b> , 157, 771-5	4	13
233	Unusual molecular findings in Kindler syndrome. <i>British Journal of Dermatology</i> , <b>2007</b> , 157, 1252-6	4	33
232	Recurrent KIND1 (C20orf42) gene mutation, c.676insC, in a Brazilian pedigree with Kindler syndrome. <i>British Journal of Dermatology</i> , <b>2007</b> , 157, 1281-4	4	16
231	Globalization of DNA-based prenatal diagnosis for recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , <b>2007</b> , 32, 687-9	1.8	7
230	The molecular basis of lipoid proteinosis: mutations in extracellular matrix protein 1. <i>Experimental Dermatology</i> , <b>2007</b> , 16, 881-90	4	117
229	Visual inspection with acetic acid as a cervical cancer test: accuracy validated using latent class analysis. <i>BMC Medical Research Methodology</i> , <b>2007</b> , 7, 36	4.7	25
228	Diseases of epidermal keratins and their linker proteins. <i>Experimental Cell Research</i> , <b>2007</b> , 313, 1995-2009.2	4.2	81
227	Stage II follow-up on a linkage scan for bipolar disorder in the Ashkenazim provides suggestive evidence for chromosome 12p and the GRIN2B gene. <i>Genetics in Medicine</i> , <b>2007</b> , 9, 745-51	8.1	28
226	Clinical and molecular dilemmas in the diagnosis of familial epidermolysis bullosa pruriginosa. <i>Journal of the American Academy of Dermatology</i> , <b>2007</b> , 56, S77-81	4.5	26
225	Rapp-Hodgkin ectodermal dysplasia syndrome: the clinical and molecular overlap with Hay-Wells syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2006</b> , 140, 887-91	2.5	14
224	Epidermolysis bullosa. <i>British Journal of Hospital Medicine (London, England: 2005)</i> , <b>2006</b> , 67, 188-91	0.8	16
223	Naegeli-Franceschetti-Jadassohn syndrome and dermatopathia pigmentosa reticularis: two allelic ectodermal dysplasias caused by dominant mutations in KRT14. <i>American Journal of Human Genetics</i> , <b>2006</b> , 79, 724-30	11	94
222	Single cell PCR amplification of microsatellites flanking the COL7A1 gene and suitability for preimplantation genetic diagnosis of Hallopeau-Siemens recessive dystrophic epidermolysis bullosa. <i>Journal of Dermatological Science</i> , <b>2006</b> , 42, 241-8	4.3	22
221	Single nucleotide polymorphism in a commonly utilized LAMB3 primer sequence: implications for mutation detection and haplotype analysis in junctional epidermolysis bullosa. <i>Journal of Dermatological Science</i> , <b>2006</b> , 44, 48-51	4.3	2
220	Ectodermal dysplasia-skin fragility syndrome resulting from a new homozygous mutation, 888delC, in the desmosomal protein plakophilin 1. <i>Journal of the American Academy of Dermatology</i> , <b>2006</b> , 55, 157-61	4.5	35

219	Phenotype, genotype, and sustained response to anakinra in 22 patients with autoinflammatory disease associated with CIAS-1/NALP3 mutations. <i>Archives of Dermatology</i> , <b>2006</b> , 142, 1591-7		138
218	Kindler syndrome: a new mutation and new diagnostic possibilities. <i>Archives of Dermatology</i> , <b>2006</b> , 142, 620-4		22
217	Extracellular matrix protein 1 inhibits the activity of matrix metalloproteinase 9 through high-affinity protein/protein interactions. <i>Experimental Dermatology</i> , <b>2006</b> , 15, 300-7	4	67
216	Prenatal diagnosis for severe inherited skin disorders: 25 years' experience. <i>British Journal of Dermatology</i> , <b>2006</b> , 154, 106-13	4	64
215	Preimplantation genetic diagnosis of skin fragility-ectodermal dysplasia syndrome. <i>British Journal of Dermatology</i> , <b>2006</b> , 154, 546-50	4	25
214	Retrospective diagnosis of Kindler syndrome in a 37-year-old man. <i>Clinical and Experimental Dermatology</i> , <b>2006</b> , 31, 45-7	1.8	14
213	Target proteins in inherited and acquired blistering skin disorders. <i>Clinical and Experimental Dermatology</i> , <b>2006</b> , 31, 252-9	1.8	30
212	Second cite. <i>Clinical and Experimental Dermatology</i> , <b>2006</b> , 31, 826-8	1.8	4
211	Molecular basis of Kindler syndrome in Italy: novel and recurrent Alu/Alu recombination, splice site, nonsense, and frameshift mutations in the KIND1 gene. <i>Journal of Investigative Dermatology</i> , <b>2006</b> , 126, 1776-83	4.3	47
210	Complete maternal isodisomy of chromosome 3 in a child with recessive dystrophic epidermolysis bullosa but no other phenotypic abnormalities. <i>Journal of Investigative Dermatology</i> , <b>2006</b> , 126, 2039-43	4.3	28
209	Bipolar I disorder and schizophrenia: a 440-single-nucleotide polymorphism screen of 64 candidate genes among Ashkenazi Jewish case-parent trios. <i>American Journal of Human Genetics</i> , <b>2005</b> , 77, 918-36	<sup>11</sup>	323
208	Accumulation of Krebs cycle intermediates and over-expression of HIF1alpha in tumours which result from germline FH and SDH mutations. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 2231-9	5.6	683
207	Extracellular matrix protein 1 interacts with the domain III of fibulin-1C and 1D variants through its central tandem repeat 2. <i>Biochemical and Biophysical Research Communications</i> , <b>2005</b> , 333, 1327-33	3.4	32
206	Three-dimensional imaging reveals major changes in skin microvasculature in lipoid proteinosis and lichen sclerosus. <i>Journal of Dermatological Science</i> , <b>2005</b> , 38, 215-24	4.3	26
205	Normal and abnormal mechanisms of gene splicing and relevance to inherited skin diseases. <i>Journal of Dermatological Science</i> , <b>2005</b> , 40, 73-84	4.3	27
204	Neonatal diagnosis of Kindler syndrome. <i>Journal of Dermatological Science</i> , <b>2005</b> , 39, 183-5	4.3	19
203	Inherited disorders of desmosomes. <i>Australasian Journal of Dermatology</i> , <b>2005</b> , 46, 221-9	1.3	57
202	Thalidomide in the management of epidermolysis bullosa pruriginosa. <i>British Journal of Dermatology</i> , <b>2005</b> , 152, 1332-4	4	44

201	Transient bullous dermolysis of the newborn in three generations. <i>British Journal of Dermatology</i> , <b>2005</b> , 153, 1058-63	4	34
200	Complete paternal uniparental isodisomy of chromosome 1 resulting in Herlitz junctional epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , <b>2005</b> , 30, 71-4	1.8	26
199	Familial multiple cutaneous and uterine leiomyomas associated with papillary renal cell cancer. <i>Clinical and Experimental Dermatology</i> , <b>2005</b> , 30, 75-8	1.8	35
198	Prenatal diagnosis of Herlitz junctional epidermolysis bullosa in nonidentical twins. <i>Clinical and Experimental Dermatology</i> , <b>2005</b> , 30, 180-2	1.8	5
197	An Indian child with Kindler syndrome resulting from a new homozygous nonsense mutation (C468X) in the KIND1 gene. <i>Clinical and Experimental Dermatology</i> , <b>2005</b> , 30, 286-8	1.8	13
196	Rapp-Hodgkin syndrome and the tail of p63. <i>Clinical and Experimental Dermatology</i> , <b>2005</b> , 30, 183-6	1.8	24
195	Molecular abnormalities of the desmosomal protein desmoplakin in human disease. <i>Clinical and Experimental Dermatology</i> , <b>2005</b> , 30, 261-6	1.8	29
194	Infantile acquired zinc deficiency resembling acrodermatitis enteropathica. <i>Clinical and Experimental Dermatology</i> , <b>2005</b> , 30, 594-5	1.8	8
193	LEKTI is localized in lamellar granules, separated from KLK5 and KLK7, and is secreted in the extracellular spaces of the superficial stratum granulosum. <i>Journal of Investigative Dermatology</i> , <b>2005</b> , 124, 360-6	4.3	123
192	Genotype-phenotype correlation in recessive dystrophic epidermolysis bullosa: when missense doesn't make sense. <i>Journal of Investigative Dermatology</i> , <b>2005</b> , 124, 863-6	4.3	23
191	Human hair abnormalities resulting from inherited desmosome gene mutations. <i>Keio Journal of Medicine</i> , <b>2005</b> , 54, 72-9	1.6	21
190	Clinical and molecular significance of splice site mutations in the plakophilin 1 gene in patients with ectodermal dysplasia-skin fragility syndrome. <i>Acta Dermato-Venereologica</i> , <b>2005</b> , 85, 386-8	2.2	8
189	Clinical and molecular abnormalities in lipid proteinosis. <i>European Journal of Dermatology</i> , <b>2005</b> , 15, 344-6	0.8	15
188	Translational benefits from research on rare genodermatoses. <i>Australasian Journal of Dermatology</i> , <b>2004</b> , 45, 89-93	1.3	8
187	Striate palmoplantar keratoderma arising from desmoplakin and desmoglein 1 mutations is associated with contrasting perturbations of desmosomes and the keratin filament network. <i>British Journal of Dermatology</i> , <b>2004</b> , 150, 878-91	4	50
186	Clinical and molecular characterization of lipid proteinosis in Namaqualand, South Africa. <i>British Journal of Dermatology</i> , <b>2004</b> , 151, 413-23	4	79
185	An Indian child with lipid proteinosis resulting from a recurrent frameshift mutation (507delT) in the extracellular matrix protein 1 gene. <i>British Journal of Dermatology</i> , <b>2004</b> , 151, 726-7	4	6
184	Characterization of IgG autoantibodies to extracellular matrix protein 1 in lichen sclerosis. <i>Clinical and Experimental Dermatology</i> , <b>2004</b> , 29, 499-504	1.8	52

183	Ectodermal dysplasia showing clinical overlap between AEC, Rapp-Hodgkin and CHAND syndromes. <i>Clinical and Experimental Dermatology</i> , <b>2004</b> , 29, 486-8	1.8	21
182	Altered expression of L-arginine metabolism pathway genes in chronic wounds in recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , <b>2004</b> , 29, 664-8	1.8	9
181	ADULT ectodermal dysplasia syndrome resulting from the missense mutation R298Q in the p63 gene. <i>Clinical and Experimental Dermatology</i> , <b>2004</b> , 29, 669-72	1.8	26
180	Increased risk of squamous cell carcinoma in junctional epidermolysis bullosa. <i>Journal of the European Academy of Dermatology and Venereology</i> , <b>2004</b> , 18, 521-6	4.6	26
179	Reduced expression of insulin-like growth factor-binding protein-3 (IGFBP-3) in Squamous cell carcinoma complicating recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>2004</b> , 122, 1302-9	4.3	20
178	Recurrent mutations in kindlin-1, a novel keratinocyte focal contact protein, in the autosomal recessive skin fragility and photosensitivity disorder, Kindler syndrome. <i>Journal of Investigative Dermatology</i> , <b>2004</b> , 122, 78-83	4.3	61
177	Keratinocyte heal thyself: a new form of "natural gene therapy". <i>Journal of Investigative Dermatology</i> , <b>2004</b> , 122, x-xi	4.3	4
176	An indirect test of the new mutation hypothesis associating advanced paternal age with the etiology of schizophrenia. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 124B, 6-9		18
175	Genomewide linkage scan for bipolar-disorder susceptibility loci among Ashkenazi Jewish families. <i>American Journal of Human Genetics</i> , <b>2004</b> , 75, 204-19	11	83
174	Rapid diagnosis of lipid proteinosis using an anti-extracellular matrix protein 1 (ECM1) antibody. <i>Journal of Dermatological Science</i> , <b>2004</b> , 35, 151-3	4.3	13
173	Molecular basis of lipid proteinosis in two Indian siblings. <i>Journal of Dermatology</i> , <b>2004</b> , 31, 764-6	1.6	8
172	Five latent factors underlying schizophrenia: analysis and relationship to illnesses in relatives. <i>Schizophrenia Bulletin</i> , <b>2004</b> , 30, 855-73	1.3	29
171	Generalized dystonia and striatal calcifications with lipid proteinosis. <i>Neurology</i> , <b>2004</b> , 63, 2168-9	6.5	24
170	Development of antigen-specific ELISA for circulating autoantibodies to extracellular matrix protein 1 in lichen sclerosus. <i>Journal of Clinical Investigation</i> , <b>2004</b> , 113, 1550-1559	15.9	38
169	Development of antigen-specific ELISA for circulating autoantibodies to extracellular matrix protein 1 in lichen sclerosus. <i>Journal of Clinical Investigation</i> , <b>2004</b> , 113, 1550-9	15.9	15
168	An unusual N-terminal deletion of the laminin alpha3a isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. <i>Human Molecular Genetics</i> , <b>2003</b> , 12, 2395-409	5.6	100
167	Lack of plakophilin 1 increases keratinocyte migration and reduces desmosome stability. <i>Journal of Cell Science</i> , <b>2003</b> , 116, 3303-14	5.3	89
166	Desmosomes exhibit site-specific features in human palm skin. <i>Experimental Dermatology</i> , <b>2003</b> , 12, 378-88		23

165	Extracellular matrix protein 1 gene (ECM1) mutations in lipid proteinosis and genotype-phenotype correlation. <i>Journal of Investigative Dermatology</i> , <b>2003</b> , 120, 345-50	4.3	93
164	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. <i>Journal of Investigative Dermatology</i> , <b>2003</b> , 121, 96-103	4.3	51
163	Dilemmas in distinguishing between dominant and recessive forms of dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>2003</b> , 149, 810-8	4	25
162	A case of lipid proteinosis with molecular diagnosis. <i>British Journal of Dermatology</i> , <b>2003</b> , 148, 1293-1293		
161	Molecular basis of lipid proteinosis in a Libyan family. <i>Clinical and Experimental Dermatology</i> , <b>2003</b> , 28, 545-8	1.8	26
160	Autoantibodies to extracellular matrix protein 1 in lichen sclerosus. <i>Lancet, The</i> , <b>2003</b> , 362, 118-23	4.0	236
159	Loss of kindlin-1, a human homolog of the <i>Caenorhabditis elegans</i> actin-extracellular-matrix linker protein UNC-112, causes Kindler syndrome. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 174-87	11	263
158	Genomewide linkage scan for schizophrenia susceptibility loci among Ashkenazi Jewish families shows evidence of linkage on chromosome 10q22. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 601-11 <sup>11</sup>		94
157	Mutations in the gene encoding capillary morphogenesis protein 2 cause juvenile hyaline fibromatosis and infantile systemic hyalinosis. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 791-800	11	180
156	Heterozygous mutation in the SAM domain of p63 underlies Rapp-Hodgkin ectodermal dysplasia. <i>Journal of Dental Research</i> , <b>2003</b> , 82, 433-7	8.1	58
155	Genotype-phenotype correlation in skin fragility-ectodermal dysplasia syndrome resulting from mutations in plakophilin 1. <i>Experimental Dermatology</i> , <b>2002</b> , 11, 107-14	4	49
154	Compound heterozygosity for non-sense and mis-sense mutations in desmoplakin underlies skin fragility/woolly hair syndrome. <i>Journal of Investigative Dermatology</i> , <b>2002</b> , 118, 232-8	4.3	109
153	Frameshift mutation in the V2 domain of human keratin 1 results in striate palmoplantar keratoderma. <i>Journal of Investigative Dermatology</i> , <b>2002</b> , 118, 838-44	4.3	83
152	Refined mapping of Naegeli-Franceschetti- Jadassohn syndrome to a 6 cM interval on chromosome 17q11.2-q21 and investigation of candidate genes. <i>Journal of Investigative Dermatology</i> , <b>2002</b> , 119, 692-8	4.3	31
151	Common mutations in Arg304 of the p63 gene in ectrodactyly, ectodermal dysplasia, clefting syndrome: lack of genotype-phenotype correlation and implications for mutation detection strategies. <i>Journal of Investigative Dermatology</i> , <b>2002</b> , 119, 1202-3	4.3	10
150	Genotype-phenotype correlation in italian patients with dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>2002</b> , 119, 1456-62	4.3	49
149	Human Ro60 (SSA2) genomic organization and sequence alterations, examined in cutaneous lupus erythematosus. <i>British Journal of Dermatology</i> , <b>2002</b> , 146, 210-5	4	9
148	EEC (Ectrodactyly, Ectodermal dysplasia, Clefting) syndrome: heterozygous mutation in the p63 gene (R279H) and DNA-based prenatal diagnosis. <i>British Journal of Dermatology</i> , <b>2002</b> , 146, 216-20	4	26

147	A recurrent mutation in the loricrin gene underlies the ichthyotic variant of Vohwinkel syndrome. <i>Clinical and Experimental Dermatology</i> , <b>2002</b> , 27, 243-6	1.8	36
146	Psoriasis bullosa acquisita. <i>Clinical and Experimental Dermatology</i> , <b>2002</b> , 27, 665-9	1.8	22
145	Lipoid proteinosis maps to 1q21 and is caused by mutations in the extracellular matrix protein 1 gene (ECM1). <i>Human Molecular Genetics</i> , <b>2002</b> , 11, 833-40	5.6	192
144	Bone metabolism in children with epidermolysis bullosa. <i>Journal of Pediatrics</i> , <b>2002</b> , 140, 467-9	3.6	29
143	Recent advances in the molecular basis of inherited skin diseases. <i>Advances in Genetics</i> , <b>2001</b> , 43, 1-32	3.3	11
142	Genomic localization, organization and amplification of the human zinc transporter protein gene, ZNT4, and exclusion as a candidate gene in different clinical variants of acrodermatitis enteropathica. <i>Archives of Dermatological Research</i> , <b>2001</b> , 293, 392-6	3.3	9
141	Alpha 6 beta 4 integrin abnormalities in junctional epidermolysis bullosa with pyloric atresia. <i>British Journal of Dermatology</i> , <b>2001</b> , 144, 408-14	4	52
140	New mutations in keratin 1 that cause bullous congenital ichthyosiform erythroderma and keratin 2e that cause ichthyosis bullosa of Siemens. <i>British Journal of Dermatology</i> , <b>2001</b> , 145, 330-5	4	24
139	Three cases of de novo dominant dystrophic epidermolysis bullosa associated with the mutation G2043R in COL7A1. <i>Clinical and Experimental Dermatology</i> , <b>2001</b> , 26, 97-9	1.8	14
138	Searching for candidate genes in the new millennium. <i>Clinical and Experimental Dermatology</i> , <b>2001</b> , 26, 279-83	1.8	5
137	Keratinocyte adhesion and the missing link: from Dowling-Meara to Hay-Wells. St John's Hospital Dermatological Society Annual Oration 2000. <i>Clinical and Experimental Dermatology</i> , <b>2001</b> , 26, 296-304	1.8	5
136	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , <b>2001</b> , 26, 93-96	1.8	3
135	A comparison of different lysis buffers to assess allele dropout from single cells for preimplantation genetic diagnosis. <i>Prenatal Diagnosis</i> , <b>2001</b> , 21, 490-7	3.2	57
134	Corneodesmosin expression in psoriasis vulgaris differs from normal skin and other inflammatory skin disorders. <i>Laboratory Investigation</i> , <b>2001</b> , 81, 969-76	5.9	58
133	Spectrum of dominant mutations in the desmosomal cadherin desmoglein 1, causing the skin disease striate palmoplantar keratoderma. <i>European Journal of Human Genetics</i> , <b>2001</b> , 9, 197-203	5.3	91
132	Hay-Wells syndrome is caused by heterozygous missense mutations in the SAM domain of p63. <i>Human Molecular Genetics</i> , <b>2001</b> , 10, 221-9	5.6	268
131	Immunogold electron microscopy using skin in Michel's medium intended for immunofluorescence analysis. <i>Clinics in Dermatology</i> , <b>2001</b> , 19, 638-41	3	3
130	Gene mutations, great expectations. <i>Clinics in Dermatology</i> , <b>2001</b> , 19, 59-64	3	2

129	Adjunctive testing for cervical cancer in low resource settings with visual inspection, HPV, and the Pap smear. <i>International Journal of Gynecology and Obstetrics</i> , <b>2001</b> , 72, 47-53	4	61
128	Dominant dystrophic epidermolysis bullosa presenting as familial nail dystrophy. <i>Clinical and Experimental Dermatology</i> , <b>2001</b> , 26, 93-96	1.8	1
127	HPV-based cervical cancer screening in a population at high risk for HIV infection. <i>International Journal of Cancer</i> , <b>2000</b> , 85, 206-210	7.5	67
126	Preimplantation genetic diagnosis of compound heterozygous mutations leading to ablation of plakophilin-1 (PKP1) and resulting in skin fragility ectodermal dysplasia syndrome: a case report. <i>Prenatal Diagnosis</i> , <b>2000</b> , 20, 1055-62	3.2	22
125	Genomic amplification of the human plakophilin 1 gene and detection of a new mutation in ectodermal dysplasia/skin fragility syndrome. <i>Journal of Investigative Dermatology</i> , <b>2000</b> , 115, 368-74	4.3	64
124	The gene for Naegeli-Franceschetti-Jadassohn syndrome maps to 17q21. <i>Journal of Investigative Dermatology</i> , <b>2000</b> , 115, 694-8	4.3	18
123	A homozygous missense mutation in the cytoplasmic tail of beta4 integrin, G931D, that disrupts hemidesmosome assembly and underlies Non-Herlitz junctional epidermolysis bullosa without pyloric atresia?. <i>Journal of Investigative Dermatology</i> , <b>2000</b> , 114, 1061-4	4.3	32
122	Heterozygous germline missense mutation in the p63 gene underlying EEC syndrome. <i>Clinical and Experimental Dermatology</i> , <b>2000</b> , 25, 441-3	1.8	33
121	The molecular basis of dystrophic epidermolysis bullosa in Mexico. <i>International Journal of Dermatology</i> , <b>2000</b> , 39, 436-42	1.7	22
120	Genomic organization and amplification of the human plakoglobin gene (JUP). <i>Experimental Dermatology</i> , <b>2000</b> , 9, 323-6	4	16
119	Evaluation of a human papillomavirus assay in cervical screening in Zimbabwe. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , <b>2000</b> , 107, 33-8	3.7	50
118	Genomic organization and amplification of the human keratin 15 and keratin 19 genes. <i>Biochemical and Biophysical Research Communications</i> , <b>2000</b> , 267, 462-5	3.4	9
117	Genomic organization and amplification of the human epidermal type II keratin genes K1 and K5. <i>Biochemical and Biophysical Research Communications</i> , <b>2000</b> , 274, 149-52	3.4	19
116	Genomic organization and amplification of the human desmosomal cadherin genes DSC1 and DSC3, encoding desmocollin types 1 and 3. <i>Biochemical and Biophysical Research Communications</i> , <b>2000</b> , 276, 454-60	3.4	8
115	Revised classification system for inherited epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , <b>2000</b> , 42, 1051-1066	4.5	333
114	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. <i>Journal of the American Academy of Dermatology</i> , <b>2000</b> , 43, 135-7	4.5	44
113	A disease-associated glycine substitution in BP180 (type XVII collagen) leads to a local destabilization of the major collagen triple helix. <i>Matrix Biology</i> , <b>2000</b> , 19, 223-33	11.4	13
112	Prenatal diagnosis for inherited skin diseases. <i>Clinics in Dermatology</i> , <b>2000</b> , 18, 643-8	3	14

111	Revised classification system for inherited epidermolysis bullosa: Report of the Second International Consensus Meeting on diagnosis and classification of epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , <b>2000</b> , 42, 1051-1066	4.5	72
110	HPV-based cervical cancer screening in a population at high risk for HIV infection <b>2000</b> , 85, 206		24
109	Moderation of phenotypic severity in dystrophic and junctional forms of epidermolysis bullosa through in-frame skipping of exons containing non-sense or frameshift mutations. <i>Journal of Investigative Dermatology</i> , <b>1999</b> , 113, 314-21	4.3	53
108	Skin fragility and hypohidrotic ectodermal dysplasia resulting from ablation of plakophilin 1. <i>British Journal of Dermatology</i> , <b>1999</b> , 140, 297-307	4	92
107	Defining target antigens in linear IgA disease using skin from subjects with inherited epidermolysis bullosa as a substrate for indirect immunofluorescence microscopy. <i>British Journal of Dermatology</i> , <b>1999</b> , 141, 475-80	4	10
106	A recurrent COL7A1 mutation, R2814X, in British patients with recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , <b>1999</b> , 24, 37-9	1.8	10
105	Frequency of the CCR5 gene 32-basepair deletion in Hispanic Mexicans. <i>Clinical and Experimental Dermatology</i> , <b>1999</b> , 24, 127-9	1.8	5
104	Recurrent molecular abnormalities in type VII collagen in Southern Italian patients with recessive dystrophic epidermolysis bullosa. <i>Clinical and Experimental Dermatology</i> , <b>1999</b> , 24, 232-5	1.8	8
103	Mutations in the translation initiation codon of the protoporphyrinogen oxidase gene underlie variegate porphyria. <i>Clinical and Experimental Dermatology</i> , <b>1999</b> , 24, 296-301	1.8	14
102	Recurrent COL7A1 mutations in Japanese patients with dystrophic epidermolysis bullosa: positional effects of premature termination codon mutations on clinical severity. Japanese Collaborative Study Group on Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , <b>1999</b> , 112, 991-3	4.3	27
101	Allelic heterogeneity of dominant and recessive COL7A1 mutations underlying epidermolysis bullosa pruriginosa. <i>Journal of Investigative Dermatology</i> , <b>1999</b> , 112, 984-7	4.3	59
100	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. <i>Journal of Investigative Dermatology</i> , <b>1999</b> , 113, 673-86	4.3	63
99	Striate palmoplantar keratoderma resulting from desmoplakin haploinsufficiency. <i>Journal of Investigative Dermatology</i> , <b>1999</b> , 113, 940-6	4.3	112
98	A recurrent frameshift mutation in exon 19 of the type VII collagen gene (COL7A1) in Mexican patients with recessive dystrophic epidermolysis bullosa. <i>Experimental Dermatology</i> , <b>1999</b> , 8, 22-9	4	7
97	The gene for hypotrichosis of Marie Unna maps between D8S258 and D8S298: exclusion of the hr gene by cDNA and genomic sequencing. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 413-9	11	45
96	Dyskeratosis congenita: new clinical and molecular insights into ribosome function. <i>Lancet, The</i> , <b>1999</b> , 353, 1204-5	4.0	6
95	Hereditary diseases of desmosomes. <i>Journal of Dermatological Science</i> , <b>1999</b> , 20, 85-91	4.3	27
94	A homozygous nonsense mutation in the zinc-finger domain of the human hairless gene underlies congenital atrichia. <i>Journal of Investigative Dermatology</i> , <b>1999</b> , 113, 281-3	4.3	31

93	A novel genodermatosis caused by mutations in plakophilin 1, a structural component of desmosomes. <i>Journal of Dermatology</i> , <b>1999</b> , 26, 764-9	1.6	20
92	Preimplantation genetic diagnosis of severe inherited skin diseases. <i>Experimental Dermatology</i> , <b>1998</b> , 7, 65-72	4	34
91	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex. <i>Human Mutation</i> , <b>1998</b> , 11, 279-85	4.7	36
90	Hemidesmosomes show abnormal association with the keratin filament network in junctional forms of epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1998</b> , 110, 132-7	4.3	50
89	Cycloheximide facilitates the identification of aberrant transcripts resulting from a novel splice-site mutation in COL17A1 in a patient with generalized atrophic benign epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1998</b> , 110, 165-9	4.3	21
88	Novel mutations in the LAMB3 gene shared by two Japanese unrelated families with Herlitz junctional epidermolysis bullosa, and their application for prenatal testing. <i>Journal of Investigative Dermatology</i> , <b>1998</b> , 110, 174-8	4.3	22
87	Homozygous variegate porphyria: identification of mutations on both alleles of the protoporphyrinogen oxidase gene in a severely affected proband. <i>Journal of Investigative Dermatology</i> , <b>1998</b> , 110, 452-5	4.3	32
86	Prognostic implications of determining 180 kDa bullous pemphigoid antigen (BPAG2) gene/protein pathology in neonatal junctional epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>1998</b> , 138, 661-6	4	10
85	Frameshift mutations in the type VII collagen gene (COL7A1) in five Mexican cousins with recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>1998</b> , 138, 852-8	4	12
84	E210K mutation in the gene encoding the beta3 chain of laminin-5 (LAMB3) is predictive of a phenotype of generalized atrophic benign epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>1998</b> , 139, 325-31	4	20
83	A recurrent glycine substitution mutation, G2043R, in the type VII collagen gene (COL7A1) in dominant dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>1998</b> , 139, 730-7	4	28
82	Pyloric atresia-junctional epidermolysis bullosa syndrome: mutations in the integrin beta4 gene (ITGB4) in two unrelated patients with mild disease. <i>British Journal of Dermatology</i> , <b>1998</b> , 139, 862-71	4	42
81	A missense mutation in the zinc-finger domain of the human hairless gene underlies congenital atrichia in a family of Irish travellers. <i>American Journal of Human Genetics</i> , <b>1998</b> , 63, 984-91	11	65
80	Alopecia universalis associated with a mutation in the human hairless gene. <i>Science</i> , <b>1998</b> , 279, 720-4	33.3	374
79	Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex <b>1998</b> , 11, 279		3
78	Molecular basis of blistering skin diseases. <i>British Journal of Hospital Medicine</i> , <b>1998</b> , 59, 28-32		
77	The role of immunohistochemistry in the diagnosis of the non-lethal forms of junctional epidermolysis bullosa. <i>Journal of Dermatological Science</i> , <b>1997</b> , 14, 68-75	4.3	20
76	Training for Norplant implant removal: assessment of learning curves and competency. <i>Obstetrics and Gynecology</i> , <b>1997</b> , 89, 174-8	4.9	13

75	The molecular pathology of progressive symmetric erythrokeratoderma: a frameshift mutation in the lorcrin gene and perturbations in the cornified cell envelope. <i>American Journal of Human Genetics</i> , <b>1997</b> , 61, 581-9	11	137
74	Detection of Novel LAMC2 Mutations in Herlitz Junctional Epidermolysis Bullosa. <i>Molecular Medicine</i> , <b>1997</b> , 3, 124-135	6.2	28
73	Lack of the R59W South African founder effect mutation in protoporphyrinogen oxidase in a British patient with homozygous variegate porphyria. <i>British Journal of Dermatology</i> , <b>1997</b> , 136, 292	4	
72	Premature termination codons are present on both alleles of the bullous pemphigoid antigen 2/type XVII collagen gene in five Austrian families with generalized atrophic benign epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1997</b> , 108, 463-8	4.3	38
71	Predominance of the recurrent mutation R635X in the LAMB3 gene in European patients with Herlitz junctional epidermolysis bullosa has implications for mutation detection strategy. <i>Journal of Investigative Dermatology</i> , <b>1997</b> , 109, 232-7	4.3	34
70	Recurrent mutations in the type VII collagen gene (COL7A1) in patients with recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1997</b> , 109, 246-9	4.3	47
69	Glycine substitution mutations in the type VII collagen gene (COL7A1) in dystrophic epidermolysis bullosa: implications for genetic counseling. <i>Journal of Investigative Dermatology</i> , <b>1997</b> , 108, 224-8	4.3	36
68	Mutations in the plakophilin 1 gene result in ectodermal dysplasia/skin fragility syndrome. <i>Nature Genetics</i> , <b>1997</b> , 17, 240-4	36.3	330
67	Absence of R42X and R635X mutations in the LAMB3 gene in 12 Japanese patients with junctional epidermolysis bullosa. <i>Archives of Dermatological Research</i> , <b>1997</b> , 289, 174-6	3.3	15
66	Immunohistochemical analysis of the skin in junctional epidermolysis bullosa using laminin 5 chain specific antibodies is of limited value in predicting the underlying gene mutation. <i>British Journal of Dermatology</i> , <b>1997</b> , 136, 817-822	4	5
65	Recessive epidermolysis bullosa simplex associated with plectin mutations: infantile respiratory complications in two unrelated cases. <i>British Journal of Dermatology</i> , <b>1997</b> , 137, 898-906	4	7
64	A recurrent laminin 5 mutation in British patients with lethal (Herlitz) junctional epidermolysis bullosa: evidence for a mutational hotspot rather than propagation of an ancestral allele. <i>British Journal of Dermatology</i> , <b>1997</b> , 136, 674-677	4	1
63	MUTATION-BASED PRENATAL DIAGNOSIS OF HERLITZ JUNCTIONAL EPIDERMOLYSIS BULLOSA <b>1997</b> , 17, 343-354		45
62	Recessive epidermolysis bullosa simplex associated with plectin mutations: infantile respiratory complications in two unrelated cases. <i>British Journal of Dermatology</i> , <b>1997</b> , 137, 898-906	4	39
61	A recurrent laminin 5 mutation in British patients with lethal (Herlitz) junctional epidermolysis bullosa: evidence for a mutational hotspot rather than propagation of an ancestral allele. <i>British Journal of Dermatology</i> , <b>1997</b> , 136, 674-677	4	12
60	Immunohistochemical analysis of the skin in junctional epidermolysis bullosa using laminin 5 chain specific antibodies is of limited value in predicting the underlying gene mutation. <i>British Journal of Dermatology</i> , <b>1997</b> , 136, 817-822	4	19
59	Cloning of the human type XVII collagen gene (COL17A1), and detection of novel mutations in generalized atrophic benign epidermolysis bullosa. <i>American Journal of Human Genetics</i> , <b>1997</b> , 60, 352-65 <sup>11</sup>		85
58	First trimester DNA-based exclusion of recessive dystrophic epidermolysis bullosa from chorionic villus sampling. <i>British Journal of Dermatology</i> , <b>1996</b> , 134, 734-9	4	21

57	A molecular defect in loricrin, the major component of the cornified cell envelope, underlies Vohwinkel's syndrome. <i>Nature Genetics</i> , <b>1996</b> , 13, 70-7	36.3	214
56	Clinicopathological correlations of compound heterozygous COL7A1 mutations in recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1996</b> , 107, 171-7	4.3	45
55	Molecular basis of recessive dystrophic epidermolysis bullosa: genotype/phenotype correlation in a case of moderate clinical severity. <i>Journal of Investigative Dermatology</i> , <b>1996</b> , 106, 119-24	4.3	65
54	Compound heterozygosity for nonsense and missense mutations in the LAMB3 gene in nonlethal junctional epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1996</b> , 106, 1157-9	4.3	26
53	Influence of the second COL7A1 mutation in determining the phenotypic severity of recessive dystrophic epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1996</b> , 106, 766-70	4.3	40
52	A homozygous deletion mutation in the gene encoding the 180-kDa bullous pemphigoid antigen (BPAG2) in a family with generalized atrophic benign epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1996</b> , 106, 771-4	4.3	60
51	A recurrent homozygous nonsense mutation within the LAMA3 gene as a cause of Herlitz junctional epidermolysis bullosa in patients of Pakistani ancestry: evidence for a founder effect. <i>Journal of Investigative Dermatology</i> , <b>1996</b> , 106, 781-4	4.3	22
50	Mutational hotspots in the LAMB3 gene in the lethal (Herlitz) type of junctional epidermolysis bullosa. <i>Human Molecular Genetics</i> , <b>1996</b> , 5, 231-7	5.6	91
49	Loss of plectin causes epidermolysis bullosa with muscular dystrophy: cDNA cloning and genomic organization. <i>Genes and Development</i> , <b>1996</b> , 10, 1724-35	12.6	238
48	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. <i>American Journal of Pathology</i> , <b>1996</b> , 148, 1787-96	5.8	83
47	Glycine substitutions in the triple-helical region of type VII collagen result in a spectrum of dystrophic epidermolysis bullosa phenotypes and patterns of inheritance. <i>American Journal of Human Genetics</i> , <b>1996</b> , 58, 671-81	11	76
46	Institutionalizing fertility management/human sexuality training in Colombian nursing schools. <i>Advances in Contraception: the Official Journal of the Society for the Advancement of Contraception</i> , <b>1995</b> , 11, 325-34		
45	Usefulness of a clinical scoring system to anticipate difficulty of Norplant removal. <i>Advances in Contraception: the Official Journal of the Society for the Advancement of Contraception</i> , <b>1995</b> , 11, 345-52		4
44	Genetic basis of lethal junctional epidermolysis bullosa in an affected fetus: implications for prenatal diagnosis in one family. <i>Prenatal Diagnosis</i> , <b>1995</b> , 15, 647-54	3.2	27
43	Serum sickness-like illness following streptokinase therapy. A case report. <i>Clinical and Experimental Dermatology</i> , <b>1995</b> , 20, 468-70	1.8	2
42	Mutations in the 180-kD bullous pemphigoid antigen (BPAG2), a hemidesmosomal transmembrane collagen (COL17A1), in generalized atrophic benign epidermolysis bullosa. <i>Nature Genetics</i> , <b>1995</b> , 11, 83-6	36.3	317
41	Altered laminin 5 expression due to mutations in the gene encoding the beta 3 chain (LAMB3) in generalized atrophic benign epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , <b>1995</b> , 104, 467-74	4.3	113
40	A homozygous nonsense mutation in the alpha 3 chain gene of laminin 5 (LAMA3) in lethal (Herlitz) junctional epidermolysis bullosa. <i>Human Molecular Genetics</i> , <b>1995</b> , 4, 959-62	5.6	156

39	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. <i>Genomics</i> , <b>1995</b> , 29, 282-4	4.3	50
38	Detection of sequence variants in the gene encoding the beta 3 chain of laminin 5 (LAMB3). <i>Human Mutation</i> , <b>1995</b> , 6, 77-84	4.7	58
37	Fibrillin immunoreactivity is associated with normal or fragmented elastic microfibrils at the dermal-epidermal junction in recessive dystrophic epidermolysis bullosa. <i>British Journal of Dermatology</i> , <b>1994</b> , 131, 465-71	4	6
36	Ultrastructural clues to genetic disorders of skin: the dermal-epidermal junction. <i>Journal of Investigative Dermatology</i> , <b>1994</b> , 103, 13S-18S	4.3	26
35	Epidermolysis bullosa pruriginosa: dystrophic epidermolysis bullosa with distinctive clinicopathological features. <i>British Journal of Dermatology</i> , <b>1994</b> , 130, 617-25	4	101
34	Ultrastructural Clues to Genetic Disorders of Skin: The Dermal-Epidermal Junction. <i>Journal of Investigative Dermatology</i> , <b>1994</b> , 103, S13-S18	4.3	30
33	Immunoelectron microscopy of skin basement membrane zone antigens: a pre-embedding method using 1-nm immunogold with silver enhancement. <i>Acta Dermato-Venereologica</i> , <b>1994</b> , 74, 197-200	2.2	1
32	Cultured keratinocyte allografts and wound healing in severe recessive dystrophic epidermolysis bullosa. <i>Journal of the American Academy of Dermatology</i> , <b>1993</b> , 29, 407-19	4.5	39
31	Epidermolysis bullosa simplex Dowling-Meara: troublesome blistering and pruritus in an adult patient. <i>Dermatology</i> , <b>1993</b> , 186, 68-71	4.4	8
30	Structural variations in anchoring fibrils in dystrophic epidermolysis bullosa: correlation with type VII collagen expression. <i>Journal of Investigative Dermatology</i> , <b>1993</b> , 100, 366-72	4.3	105
29	Risk factors in schizophrenia. Season of birth, gender, and familial risk. <i>British Journal of Psychiatry</i> , <b>1992</b> , 160, 65-71	5.4	47
28	Season of birth of siblings of schizophrenic patients. <i>British Journal of Psychiatry</i> , <b>1992</b> , 160, 71-5	5.4	25
27	Oral and gastrointestinal manifestations of epidermolysis bullosa. <i>Lancet, The</i> , <b>1992</b> , 340, 1505-6	4.0	47
26	Selective involvement of keratins K1 and K10 in the cytoskeletal abnormality of epidermolytic hyperkeratosis (bullous congenital ichthyosiform erythroderma). <i>Journal of Investigative Dermatology</i> , <b>1992</b> , 99, 19-26	4.3	109
25	Mitten deformity in severe generalized recessive dystrophic epidermolysis bullosa: histological, immunofluorescence, and ultrastructural study. <i>Journal of Cutaneous Pathology</i> , <b>1992</b> , 19, 385-9	1.7	9
24	Epidermolysis bullosa complicated by squamous cell carcinoma: report of 10 cases. <i>Journal of Cutaneous Pathology</i> , <b>1992</b> , 19, 116-23	1.7	110
23	Intracellular expression of type VII collagen during wound healing in severe recessive dystrophic epidermolysis bullosa and normal human skin. <i>British Journal of Dermatology</i> , <b>1992</b> , 127, 312-7	4	22
22	Epidermolysis bullosa simplex (Dowling-Meara). A clinicopathological review. <i>British Journal of Dermatology</i> , <b>1992</b> , 126, 421-30	4	50

21	Expression of mutant p53 gene in squamous carcinoma arising in patients with recessive dystrophic epidermolysis bullosa. <i>Histopathology</i> , <b>1992</b> , 20, 237-41	7.3	34
20	Schizophrenia: gender and familial risk. <i>Journal of Psychiatric Research</i> , <b>1992</b> , 26, 17-27	5.2	46
19	Metastatic squamous cell carcinoma resembling angiosarcoma complicating dystrophic epidermolysis bullosa. <i>Dermatology</i> , <b>1991</b> , 182, 235-8	4.4	25
18	The phenotypic heterogeneity of bullous ichthyosis--a case report of three family members. <i>Clinical and Experimental Dermatology</i> , <b>1991</b> , 16, 25-7	1.8	4
17	Split skin grafting and bullous pemphigoid. <i>Clinical and Experimental Dermatology</i> , <b>1991</b> , 16, 72-3	1.8	18
16	Epidermolysis bullosa simplex (Dowling-Meara type) is a genetic disease characterized by an abnormal keratin-filament network involving keratins K5 and K14. <i>Journal of Investigative Dermatology</i> , <b>1991</b> , 97, 959-68	4.3	173
15	Aquagenic pruritus and myelodysplastic syndrome. <i>American Journal of Hematology</i> , <b>1991</b> , 37, 63	7.1	4
14	(2) Dowling-Meara epidermolysis bullosa simplex. <i>British Journal of Dermatology</i> , <b>1991</b> , 125, 44-45	4	2
13	Schizophrenia: age at onset, gender and familial risk. <i>Acta Psychiatrica Scandinavica</i> , <b>1990</b> , 82, 344-51	6.5	59
12	The presence of autoantibody to recombinant lipocortin-I in patients with psoriasis and psoriatic arthritis. <i>British Journal of Dermatology</i> , <b>1990</b> , 123, 569-72	4	8
11	Aquagenic pruritus and the myelodysplastic syndrome. <i>British Journal of Dermatology</i> , <b>1990</b> , 123, 414-5	4	28
10	Age-incidence artifacts do not account for the season-of-birth effect in schizophrenia. <i>Schizophrenia Bulletin</i> , <b>1990</b> , 16, 13-5; discussion 17-28	1.3	25
9	The power of analysis: statistical perspectives. Part 1. <i>Psychiatry Research</i> , <b>1988</b> , 23, 295-9	9.9	12
8	The Dermal-Epidermal Basement Membrane Zone in Cutaneous Wound Healing <b>1988</b> , 513-560		
7	Anatomy and Organization of Human Skin45-128		38
6	A Real-world Standard for radical prostatectomy: Analysis of the British Association of Urological Surgeons Complex Operations Reports, 2016-2018. <i>Journal of Clinical Urology</i> ,205141582110639	0.2	
5	Re-evaluating the diagnostic efficacy of PSA as a referral test to detect clinically significant prostate cancer in contemporary MRI-based image-guided biopsy pathways. <i>Journal of Clinical Urology</i> ,205141582110590	0.2	0
4	Prenatal Diagnosis of Genetic Skin Disease1-11		1

3 Prenatal Diagnosis of Inherited Skin Disorders 139.1-139.12

2 Setting standards for cystectomy using the British Association of Urological Surgeons Complex Operations Reports, 2016-2018. *Journal of Clinical Urology*, 205141582110334

0.2

1 Contemporary standards in UK nephrectomy practice: Analysis of the British Association of Urological Surgeons Complex Operations Reports, 2016-2018. *Journal of Clinical Urology*, 205141582110596