

Jouni Uitto

List of Publications by Year in Descending Order

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

827
papers

38,650
citations

100
h-index

148
g-index

886
ext. papers

42,173
ext. citations

4.8
avg, IF

7.05
L-index

#	Paper	IF	Citations
827	Novel splice mutation in <i>cdsn</i> gene causing type B peeling skin syndrome.. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2022 ,	4.6	0
826	Inhibition of the DNA Damage Response Attenuates Ectopic Calcification in Pseudoxanthoma Elasticum.. <i>Journal of Investigative Dermatology</i> , 2022 ,	4.3	2
825	Inorganic pyrophosphate deficiency syndromes and potential treatments for pathologic tissue calcification.. <i>American Journal of Pathology</i> , 2022 ,	5.8	5
824	Losartan treatment improves recessive dystrophic epidermolysis bullosa: A case series.. <i>Dermatologic Therapy</i> , 2022 , e15515	2.2	0
823	Comment on "Clinical practice guidelines for pseudoxanthoma elasticum (2017)": The importance of mutation analysis.. <i>Journal of Dermatology</i> , 2022 ,	1.6	
822	ENPP1 variants in patients with GACI and PXE expand the clinical and genetic heterogeneity of heritable disorders of ectopic calcification.. <i>PLoS Genetics</i> , 2022 , 18, e1010192	6	3
821	Genetic heterogeneity of heritable ectopic mineralization disorders in a large international cohort.. <i>Genetics in Medicine</i> , 2021 ,	8.1	2
820	Oral supplementation of inorganic pyrophosphate in pseudoxanthoma elasticum. <i>Experimental Dermatology</i> , 2021 ,	4	4
819	Systemic CXCL10 is a predictive biomarker of vitiligo lesional skin infiltration, PUVA, NB-UVB and corticosteroid treatment response and outcome. <i>Archives of Dermatological Research</i> , 2021 , 1	3.3	4
818	Dynamics and Emerging Trends in Genodermatology: A Scientometric Analysis. <i>International Journal of Dermatology and Venereology</i> , 2021 , 4, 67-69	0.5	
817	Kindler epidermolysis bullosa-like skin phenotype and downregulated basement membrane zone gene expression in poikiloderma with neutropenia and a homozygous <i>USB1</i> mutation. <i>Matrix Biology</i> , 2021 , 99, 43-57	11.4	0
816	Homozygous <i>ITGA3</i> Missense Mutation in Adults in a Family with Syndromic Epidermolysis Bullosa (ILNEB) without Pulmonary Involvement. <i>Journal of Investigative Dermatology</i> , 2021 , 141, 2752-2756	4.3	3
815	Whole-Transcriptome Analysis by RNA Sequencing for Genetic Diagnosis of Mendelian Skin Disorders in the Context of Consanguinity. <i>Clinical Chemistry</i> , 2021 , 67, 876-888	5.5	4
814	Genetic Predisposition to Numerous Large Ulcerating Basal Cell Carcinomas and Response to Immune Therapy. <i>International Journal of Dermatology and Venereology</i> , 2021 , 4, 70-75	0.5	2
813	The utility of dermal fibroblasts in treatment of skin disorders: A paradigm of recessive dystrophic epidermolysis bullosa. <i>Dermatologic Therapy</i> , 2021 , 34, e15028	2.2	0
812	Interpretation of genomic sequence variants in heritable skin diseases: A primer for clinicians. <i>Journal of the American Academy of Dermatology</i> , 2021 ,	4.5	1
811	Knockdown of <i>SDR9C7</i> Impairs Epidermal Barrier Function. <i>Journal of Investigative Dermatology</i> , 2021 , 141, 1754-1764.e1	4.3	2

810	Humans with inherited T cell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. <i>Cell</i> , 2021 , 184, 3812-3828.e30	56.2	18
809	Heterogeneous addiction to transforming growth factor-beta signalling in recessive dystrophic epidermolysis bullosa-associated cutaneous squamous cell carcinoma. <i>British Journal of Dermatology</i> , 2021 , 184, 697-708	4	3
808	GGCX mutations in a patient with overlapping pseudoxanthoma elasticum/cutis laxa-like phenotype. <i>British Journal of Dermatology</i> , 2021 , 184, 1170-1174	4	0
807	Keloid disorder: Fibroblast differentiation and gene expression profile in fibrotic skin diseases. <i>Experimental Dermatology</i> , 2021 , 30, 132-145	4	10
806	Losartan for treatment of epidermolysis bullosa: A new perspective. <i>Dermatologic Therapy</i> , 2021 , 34, e14638	2.2	2
805	Molecular Genetics and Modifier Genes in Pseudoxanthoma Elasticum, a Heritable Multisystem Ectopic Mineralization Disorder. <i>Journal of Investigative Dermatology</i> , 2021 , 141, 1148-1156	4.3	10
804	Pioneers in Dermatology and Venereology: an Interview with Professor Jouni Uitto. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2021 , 35, 569-571	4.6	
803	A phytic acid analogue INS-3001 prevents ectopic calcification in an Abcc6 mouse model of pseudoxanthoma elasticum. <i>Experimental Dermatology</i> , 2021 , 30, 853-858	4	1
802	Development of the BioHybrid Assay: Combining Primary Human Vascular Smooth Muscle Cells and Blood to Measure Vascular Calcification Propensity. <i>Cells</i> , 2021 , 10,	7.9	1
801	Ancestral patterns of recessive dystrophic epidermolysis bullosa mutations in Hispanic populations suggest sephardic ancestry. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3390-3400	2.5	
800	Functional Assessment of Missense Variants in the ABCC6 Gene Implicated in Pseudoxanthoma Elasticum, a Heritable Ectopic Mineralization Disorder. <i>Journal of Investigative Dermatology</i> , 2021 ,	4.3	2
799	Lack of efficacy of dupilumab in the treatment of keloid disorder. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2021 ,	4.6	2
798	Very-Early-Onset Inflammatory Bowel Disease in a Patient With Junctional Epidermolysis Bullosa With a Homozygous Mutation in the β Integrin Gene (ITGA6). <i>Inflammatory Bowel Diseases</i> , 2021 , 27, 1865-1869	4.5	
797	Novel PTCH1 and concurrent TP53 mutations in four patients with numerous non-syndromic basal cell carcinomas: The paradigm of oncogenic synergy. <i>Experimental Dermatology</i> , 2021 ,	4	1
796	Arrhythmogenic right ventricular cardiomyopathy in patients with biallelic JUP-associated skin fragility. <i>Scientific Reports</i> , 2020 , 10, 21622	4.9	3
795	Research Techniques Made Simple: Whole-Transcriptome Sequencing by RNA-Seq for Diagnosis of Monogenic Disorders. <i>Journal of Investigative Dermatology</i> , 2020 , 140, 1117-1126.e1	4.3	15
794	Homozygous IL1RN Mutation in Siblings with Deficiency of Interleukin-1 Receptor Antagonist (DIRA). <i>Journal of Clinical Immunology</i> , 2020 , 40, 637-642	5.7	2
793	Overview on Keloid Disorder: Phenotypic Spectrum, Connective Tissue Pathology, and Treatment Development. <i>International Journal of Dermatology and Venereology</i> , 2020 , 3, 97-103	0.5	0

792	Association of MTHFR C677T polymorphism with elevated homocysteine level and disease development in vitiligo. <i>International Journal of Immunogenetics</i> , 2020 , 47, 342-350	2.3	7
791	Genomics-based treatment in a patient with two overlapping heritable skin disorders: Epidermolysis bullosa and acrodermatitis enteropathica. <i>Human Mutation</i> , 2020 , 41, 906-912	4.7	7
790	The matriptase-prostasin proteolytic cascade in dermatologic diseases. <i>Experimental Dermatology</i> , 2020 , 29, 580-587	4	3
789	Molecular Therapeutics in Development for Epidermolysis Bullosa: Update 2020. <i>Molecular Diagnosis and Therapy</i> , 2020 , 24, 299-309	4.5	26
788	Kidney Stones are Prevalent in Individuals with Pseudoxanthoma Elasticum, a Genetic Ectopic Mineralization Disorder.. <i>International Journal of Dermatology and Venereology</i> , 2020 , 3, 198-204	0.5	1
787	Molecular Genetics of Keratinization Disorders - What's New About Ichthyosis. <i>Acta Dermato-Venereologica</i> , 2020 , 100, adv00095	2.2	9
786	Therapeutics Development for Pseudoxanthoma Elasticum and Related Ectopic Mineralization Disorders: Update 2020. <i>Journal of Clinical Medicine</i> , 2020 , 10,	5.1	10
785	Meeting Report: The First Global Congress on Epidermolysis Bullosa, EB2020 London: Toward Treatment and Cure. <i>Journal of Investigative Dermatology</i> , 2020 , 140, 1681-1687	4.3	8
784	Clinical Challenge and Call for Research on Keloid Disorder: Meeting Report from The 3rd International Keloid Research Foundation Symposium, Beijing 2019. <i>Journal of Investigative Dermatology</i> , 2020 , 140, 515-518	4.3	4
783	Meeting Report of the 4th Annual Meeting of the Chinese Society for Investigative Dermatology: Reflections on the Rise of Cutaneous Biology Research in China. <i>Journal of Investigative Dermatology</i> , 2020 , 140, 729-732.e4	4.3	1
782	Atherogenic Diet Accelerates Ectopic Mineralization in a Mouse Model of Pseudoxanthoma Elasticum. <i>International Journal of Dermatology and Venereology</i> , 2020 , 3, 91-96	0.5	1
781	Increased level of cathelicidin (LL-37) in vitiligo: Possible pathway independent from vitamin D receptor gene polymorphism. <i>Experimental Dermatology</i> , 2020 , 29, 1176-1185	4	2
780	Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. <i>British Journal of Dermatology</i> , 2020 , 183, 614-627	4	179
779	Multidisciplinary care of epidermolysis bullosa during the COVID-19 pandemic-Consensus: Recommendations by an international panel of experts. <i>Journal of the American Academy of Dermatology</i> , 2020 , 83, 1222-1224	4.5	5
778	Aberrant recruitment of leukocytes defines poor wound healing in patients with recessive dystrophic epidermolysis bullosa. <i>Journal of Dermatological Science</i> , 2020 , 100, 209-216	4.3	3
777	Coronavirus disease 2019 and epidermolysis bullosa: Report of three cases. <i>Dermatologic Therapy</i> , 2020 , 33, e14194	2.2	3
776	Keratitis-ichthyosis-deafness syndrome: Phenotypic heterogeneity and treatment perspective of patients with p.Asp50Asn GJB2 mutation. <i>Dermatologic Therapy</i> , 2020 , 33, e14493	2.2	2
775	Mycophenolate mofetil treatment of an H syndrome patient with a SLC29A3 mutation. <i>Dermatologic Therapy</i> , 2020 , 33, e14375	2.2	2

774	Genotype-phenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. <i>British Journal of Dermatology</i> , 2020 , 182, 729-737	4	19
773	Epidermolysis bullosa: diagnostic guidelines in the laboratory setting. <i>British Journal of Dermatology</i> , 2020 , 182, 526-527	4	1
772	Linear basal cell nevus with a novel mosaic PTCH1 mutation. <i>Experimental Dermatology</i> , 2020 , 29, 531-534	4	1
771	Widespread aplasia cutis congenita in sibs with PLEC1 and ITGB4 variants. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 1547-1555	2.5	2
770	Marcel F. Jonkman, MD, PhD (1957-2019). <i>Journal of Investigative Dermatology</i> , 2019 , 139, 982-983	4.3	
769	Reply to Van Gils and Vanakker. <i>Journal of Investigative Dermatology</i> , 2019 , 139, 1600-1601	4.3	
768	Inherited non-alcoholic fatty liver disease and dyslipidemia due to monoallelic ABHD5 mutations. <i>Journal of Hepatology</i> , 2019 , 71, 366-370	13.4	29
767	Adenovirus-Mediated ABCC6 Gene Therapy for Heritable Ectopic Mineralization Disorders. <i>Journal of Investigative Dermatology</i> , 2019 , 139, 1254-1263	4.3	11
766	A CIB1 Splice-Site Founder Mutation in Families with Typical Epidermodysplasia Verruciformis. <i>Journal of Investigative Dermatology</i> , 2019 , 139, 1195-1198	4.3	10
765	Biallelic KRT5 mutations in autosomal recessive epidermolysis bullosa simplex, including a complete human keratin 5 "knock-out". <i>Matrix Biology</i> , 2019 , 83, 48-59	11.4	8
764	Assessment of the risk and characterization of non-melanoma skin cancer in Kindler syndrome: study of a series of 91 patients. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 183	4.2	9
763	Quantitative Trait Locus and Integrative Genomics Revealed Candidate Modifier Genes for Ectopic Mineralization in Mouse Models of Pseudoxanthoma Elasticum. <i>Journal of Investigative Dermatology</i> , 2019 , 139, 2447-2457.e7	4.3	12
762	Hypotrichosis with juvenile macular dystrophy: Combination of whole-genome sequencing and genome-wide homozygosity mapping identifies a large deletion in CDH3 initially undetected by whole-exome sequencing-A lesson from next-generation sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e975	2.3	5
761	Applications of Spherical Nucleic Acid Nanoparticles as Delivery Systems. <i>Trends in Molecular Medicine</i> , 2019 , 25, 1066-1079	11.5	28
760	Inherited Interleukin 2-Inducible T-Cell (ITK) Kinase Deficiency in Siblings With Epidermodysplasia Verruciformis and Hodgkin Lymphoma. <i>Clinical Infectious Diseases</i> , 2019 , 68, 1938-1941	11.6	15
759	Toward treatment and cure of epidermolysis bullosa. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019 ,	11.5	12
758	Pseudoxanthoma elasticum: Dermoscopy and mutation analysis. <i>Australasian Journal of Dermatology</i> , 2019 , 60, e156-e158	1.3	1
757	Phenotypic Spectrum of Epidermolysis Bullosa: The Paradigm of Syndromic versus Non-Syndromic Skin Fragility Disorders. <i>Journal of Investigative Dermatology</i> , 2019 , 139, 522-527	4.3	30

756	Autosomal recessive congenital ichthyosis: Genomic landscape and phenotypic spectrum in a cohort of 125 consanguineous families. <i>Human Mutation</i> , 2019 , 40, 288-298	4.7	21
755	Pseudoxanthoma Elasticum as a Paradigm of Heritable Ectopic Mineralization Disorders: Pathomechanisms and Treatment Development. <i>American Journal of Pathology</i> , 2019 , 189, 216-225	5.8	37
754	A novel autosomal recessive GJB2-associated disorder: Ichthyosis follicularis, bilateral severe sensorineural hearing loss, and punctate palmoplantar keratoderma. <i>Human Mutation</i> , 2019 , 40, 217-229	4.7	6
753	Inhibition of Tissue-Nonspecific Alkaline Phosphatase Attenuates Ectopic Mineralization in the Abcc6 Mouse Model of PXE but Not in the Enpp1 Mutant Mouse Models of GACI. <i>Journal of Investigative Dermatology</i> , 2019 , 139, 360-368	4.3	31
752	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> , 2019 , 81, 91-106	11.4	28
751	Epidermodysplasia Verruciformis: Genetic Heterogeneity and EVER1 and EVER2 Mutations Revealed by Genome-Wide Analysis. <i>Journal of Investigative Dermatology</i> , 2019 , 139, 241-244	4.3	11
750	Trauma-Induced Nanohydroxyapatite Deposition in Skeletal Muscle is Sufficient to Drive Heterotopic Ossification. <i>Calcified Tissue International</i> , 2019 , 104, 411-425	3.9	8
749	Pachyonychia congenita: a case report of a successful treatment with rosuvastatin in a patient with a KRT6A mutation. <i>British Journal of Dermatology</i> , 2019 , 181, 584-586	4	12
748	Genome-wide single nucleotide polymorphism-based autozygosity mapping facilitates identification of mutations in consanguineous families with epidermolysis bullosa. <i>Experimental Dermatology</i> , 2019 , 28, 1118-1121	4	13
747	Seven novel COL7A1 mutations identified in patients with recessive dystrophic epidermolysis bullosa from Mexico. <i>Clinical and Experimental Dermatology</i> , 2018 , 43, 579-584	1.8	4
746	Epidermolysis bullosa: Molecular pathology of connective tissue components in the cutaneous basement membrane zone. <i>Matrix Biology</i> , 2018 , 71-72, 313-329	11.4	53
745	EB2017-Progress in Epidermolysis Bullosa Research toward Treatment and Cure. <i>Journal of Investigative Dermatology</i> , 2018 , 138, 1010-1016	4.3	30
744	First report of COL7A1 mutations in two patients with recessive dystrophic epidermolysis bullosa from Peru. <i>Clinical and Experimental Dermatology</i> , 2018 , 43, 719-722	1.8	1
743	The Conundrum of Allogeneic Bone Marrow Transplantation for Epidermolysis Bullosa. <i>Journal of Investigative Dermatology</i> , 2018 , 138, 1029-1031	4.3	1
742	Erythrokeratoderma: a manifestation associated with multiple types of ichthyoses with different gene defects. <i>British Journal of Dermatology</i> , 2018 , 178, e219-e221	4	3
741	The human CIB1-EVER1-EVER2 complex governs keratinocyte-intrinsic immunity to Epapillomaviruses. <i>Journal of Experimental Medicine</i> , 2018 , 215, 2289-2310	16.6	56
740	The genetic basis of hyaline fibromatosis syndrome in patients from a consanguineous background: a case series. <i>BMC Medical Genetics</i> , 2018 , 19, 87	2.1	3
739	Epidermodysplasia Verruciformis: Inborn Errors of Immunity to Human Beta-Papillomaviruses. <i>Frontiers in Microbiology</i> , 2018 , 9, 1222	5.7	32

738	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> , 2018 , 39, 1349-1354	4.7	19
737	Research Techniques Made Simple: Genome-Wide Homozygosity/Autozygosity Mapping Is a Powerful Tool for Identifying Candidate Genes in Autosomal Recessive Genetic Diseases. <i>Journal of Investigative Dermatology</i> , 2018 , 138, 1893-1900	4.3	24
736	Etidronate prevents, but does not reverse, ectopic mineralization in a mouse model of pseudoxanthoma elasticum (. <i>Oncotarget</i> , 2018 , 9, 30721-30730	3.3	20
735	Recessive mutation in tetraspanin CD151 causes Kindler syndrome-like epidermolysis bullosa with multi-systemic manifestations including nephropathy. <i>Matrix Biology</i> , 2018 , 66, 22-33	11.4	39
734	Heritable Ectopic Mineralization Disorders: Pathomechanisms and Potential Treatment. <i>Journal of Investigative Dermatology Symposium Proceedings</i> , 2018 , 19, S106-S107	1.1	1
733	Precision Medicine for Heritable Skin Diseases-The Paradigm of Epidermolysis Bullosa. <i>Journal of Investigative Dermatology Symposium Proceedings</i> , 2018 , 19, S74-S76	1.1	2
732	Zebrafish Models of Ectopic Mineralization-The Paradigm of Pseudoxanthoma Elasticum. <i>Journal of Investigative Dermatology</i> , 2018 , 138, 2301-2304	4.3	4
731	Filaggrin 2 Deficiency Results in Abnormal Cell-Cell Adhesion in the Cornified Cell Layers and Causes Peeling Skin Syndrome Type A. <i>Journal of Investigative Dermatology</i> , 2018 , 138, 1736-1743	4.3	23
730	Abcc6 Knockout Rat Model Highlights the Role of Liver in PPI Homeostasis in Pseudoxanthoma Elasticum. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 1025-1032	4.3	36
729	Misbalanced CXCL12 and CCL5 Chemotactic Signals in Vitiligo Onset and Progression. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 1126-1134	4.3	33
728	Gene-Targeted Next-Generation Sequencing Identifies a Novel CLDN1 Mutation in a Consanguineous Family With NISCH Syndrome. <i>American Journal of Gastroenterology</i> , 2017 , 112, 396-398	8.7	6
727	Phenotypic spectrum of autosomal recessive congenital ichthyosis due to PNPLA1 mutation. <i>British Journal of Dermatology</i> , 2017 , 177, 319-322	4	11
726	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> , 2017 , 137, 1842-1849	4.3	52
725	Insights into Pathomechanisms and Treatment Development in Heritable Ectopic Mineralization Disorders: Summary of the PXE International Biennial Research Symposium-2016. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 790-795	4.3	25
724	A novel mutation in ST14 at a functionally significant amino acid residue expands the spectrum of ichthyosis-hypotrichosis syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2017 , 12, 176	4.2	5
723	Hyaline Fibromatosis Syndrome: A Novel Mutation and Recurrent Founder Mutation in the CMG2/ANTXR2 Gene. <i>Acta Dermato-Venereologica</i> , 2017 , 97, 108-109	2.2	8
722	Multigene Next-Generation Sequencing Panel Identifies Pathogenic Variants in Patients with Unknown Subtype of Epidermolysis Bullosa: Subclassification with Prognostic Implications. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 2649-2652	4.3	23
721	Expanding mutation landscape and phenotypic spectrum of autosomal recessive congenital ichthyosis. <i>British Journal of Dermatology</i> , 2017 , 177, 342-343	4	

720	Autosomal recessive congenital ichthyosis: CERS3 mutations identified by a next generation sequencing panel targeting ichthyosis genes. <i>European Journal of Human Genetics</i> , 2017 , 25, 1282-1285	5.3	14
719	Pro-Inflammatory Chemokines and Cytokines Dominate the Blister Fluid Molecular Signature in Patients with Epidermolysis Bullosa and Affect Leukocyte and Stem Cell Migration. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 2298-2308	4.3	15
718	Plasma PPI Deficiency Is the Major, but Not the Exclusive, Cause of Ectopic Mineralization in an Abcc6 Mouse Model of PXE. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 2336-2343	4.3	33
717	Molecular Dynamics Simulation of the Consequences of a PYCR1 Mutation (p.Ala189Val) in Patients with Complex Connective Tissue Disorder and Severe Intellectual Disability. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 525-528	4.3	4
716	Calpain 12 Function Revealed through the Study of an Atypical Case of Autosomal Recessive Congenital Ichthyosis. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 385-393	4.3	14
715	Dystrophic Epidermolysis Bullosa: COL7A1 Mutation Landscape in a Multi-Ethnic Cohort of 152 Extended Families with High Degree of Customary Consanguineous Marriages. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 660-669	4.3	34
714	Molecular pathology of the basement membrane zone in heritable blistering diseases:: The paradigm of epidermolysis bullosa. <i>Matrix Biology</i> , 2017 , 57-58, 76-85	11.4	44
713	Gene-Targeted Next Generation Sequencing Identifies PNPLA1 Mutations in Patients with a Phenotypic Spectrum of Autosomal Recessive Congenital Ichthyosis: The Impact of Consanguinity. <i>Journal of Investigative Dermatology</i> , 2017 , 137, 678-685	4.3	21
712	SVEP1 plays a crucial role in epidermal differentiation. <i>Experimental Dermatology</i> , 2017 , 26, 423-430	4	6
711	Elevated dietary magnesium during pregnancy and postnatal life prevents ectopic mineralization in Enpp1asj mice, a model for generalized arterial calcification of infancy. <i>Oncotarget</i> , 2017 , 8, 38152-38160	3.3	10
710	Kindler syndrome, an orphan disease of cell/matrix adhesion in the skin –molecular genetics and therapeutic opportunities. <i>Expert Opinion on Orphan Drugs</i> , 2016 , 4, 845-854	1.1	4
709	Stem Cell Therapy for Epidermolysis Bullosa-Does It Work?. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 2119-2121	4.3	11
708	KRT5 and KRT14 Mutations in Epidermolysis Bullosa Simplex with Phenotypic Heterogeneity, and Evidence of Semidominant Inheritance in a Multiplex Family. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 1897-1901	4.3	14
707	Type VII Collagen Replacement Therapy in Recessive Dystrophic Epidermolysis Bullosa-How Much, How Often?. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 1079-1081	4.3	4
706	Highly branched poly(L-amino ester)s for skin gene therapy. <i>Journal of Controlled Release</i> , 2016 , 244, 336-346	11.7	78
705	Introduction to mini-review cluster on fibrotic diseases: A Festschrift to Joel Rosenbloom, M.D., Ph.D. <i>Matrix Biology</i> , 2016 , 51, 5-6	11.4	
704	Progress toward Treatment and Cure of Epidermolysis Bullosa: Summary of the DEBRA International Research Symposium EB2015. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 352-358	4.3	48
703	Keloids: The paradigm of skin fibrosis - Pathomechanisms and treatment. <i>Matrix Biology</i> , 2016 , 51, 37-46	11.4	140

702	Epidemiologic, Clinicopathologic, Diagnostic, and Management Challenges of Pityriasis Rubra Pilaris: A Case Series of 100 Patients. <i>JAMA Dermatology</i> , 2016 , 152, 670-5	5.1	37
701	Research Progress in Pseudoxanthoma Elasticum and Related Ectopic Mineralization Disorders. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 550-556	4.3	26
700	Keloids: Animal models and pathologic equivalents to study tissue fibrosis. <i>Matrix Biology</i> , 2016 , 51, 47-54	4.4	29
699	Molecular Genetics of the PI3K-AKT-mTOR Pathway in Genodermatoses: Diagnostic Implications and Treatment Opportunities. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 15-23	4.3	25
698	Dual Effects of Bisphosphonates on Ectopic Skin and Vascular Soft Tissue Mineralization versus Bone Microarchitecture in a Mouse Model of Generalized Arterial Calcification of Infancy. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 275-283	4.3	31
697	Ectopic mineralization of cartilage and collagen-rich tendons and ligaments in Enpp1asj-2J mice. <i>Oncotarget</i> , 2016 , 7, 12000-9	3.3	9
696	Variable patterns of ectopic mineralization in Enpp1asj-2J mice, a model for generalized arterial calcification of infancy. <i>Oncotarget</i> , 2016 , 7, 83837-83842	3.3	2
695	Phenotypic heterogeneity in PIK3CA-related overgrowth spectrum. <i>British Journal of Dermatology</i> , 2016 , 175, 810-4	4	8
694	Expanding genetics and phenotypic spectrum of epidermodysplasia verruciformis. <i>British Journal of Dermatology</i> , 2016 , 175, 1138-1139	4	2
693	Whole Genome Linkage Analysis Followed by Whole Exome Sequencing Identifies Nicastrin (NCSTN) as a Causative Gene in a Multiplex Family with Ectopic Mineralization Spectrum of Autoinflammatory Skin Phenotypes. <i>Journal of Investigative Dermatology</i> , 2016 , 136, 1283-1286	4.3	9
692	Mouse genome-wide association study identifies polymorphisms on chromosomes 4, 11, and 15 for age-related cardiac fibrosis. <i>Mammalian Genome</i> , 2016 , 27, 179-90	3.2	9
691	Co-existence of phenylketonuria either with maple syrup urine disease or Sandhoff disease in two patients from Iran: emphasizing the role of consanguinity. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016 , 29, 1215-1219	1.6	5
690	Chemotaxis-driven disease-site targeting of therapeutic adult stem cells in dystrophic epidermolysis bullosa. <i>Stem Cell Research and Therapy</i> , 2016 , 7, 124	8.3	6
689	Klippel-Trenaunay syndrome belongs to the PIK3CA-related overgrowth spectrum (PROS). <i>Experimental Dermatology</i> , 2016 , 25, 17-9	4	95
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