Peter C Van Den Akker

List of Publications by Year
in descending order

44 papers	1,635 citations	331259 21 h-index	301761 39 g-index
46 all docs	46 docs citations	46 times ranked	3271 citing authors

#	Article	IF	Citations
1	Loss-of-function variants in the filaggrin gene are a significant risk factor for peanut allergy. Journal of Allergy and Clinical Immunology, 2011, 127, 661-667.	1.5	424
2	Improving the diagnostic yield of exome- sequencing by predicting geneâ€“phenotype associations using large-scale gene expression analysis. Nature Communications, 2019, 10, 2837.	5.8	107
3	Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. British Journal of Dermatology, 2020, 182, 574-592.	1.4	88
4	WRN mutations in Werner syndrome patients: genomic rearrangements, unusual intronic mutations and ethnic-specific alterations. Human Genetics, 2010, 128, 103-111.	1.8	87
5	The international dystrophic epidermolysis bullosa patient registry: An online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. Human Mutation, 2011, 32, 1100-1107.	1.1	74
6	An Overview and Online Registry of Microvillus Inclusion Disease Patients and theirMYO5BMutations. Human Mutation, 2013, 34, 1597-1605.	1.1	62
7	Antisense Oligonucleotide-mediated Exon Skipping as a Systemic Therapeutic Approach for Recessive Dystrophic Epidermolysis Bullosa. Molecular Therapy - Nucleic Acids, 2016, 5, e379.	2.3	59
8	Left ventricular outflow tract obstruction: should cardiac screening be offered to first-degree relatives?. Heart, 2011, 97, 1228-1232.	1.2	56
9	New ZMPSTE24 (FACE1) mutations in patients affected with restrictive dermopathy or related progeroid syndromes and mutation update. European Journal of Human Genetics, 2014, 22, 1002-1011.	1.4	51
10	Lamin A/C -Related Cardiac Disease. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	51
11	Analysis of the functional consequences of targeted exon deletion in COL7A1 reveals prospects for dystrophic epidermolysis bullosa therapy. Molecular Therapy, 2016, 24, 1302-1311.	3.7	42
12	Mechanisms of Natural Gene Therapy in Dystrophic Epidermolysis Bullosa. Journal of Investigative Dermatology, 2014, 134, 2097-2104.	0.3	40
13	Novel insights into the epidemiology of epidermolysis bullosa (EB) from the Dutch EB Registry: EB more common than previously assumed?. Journal of the European Academy of Dermatology and Venereology, 2021, 35, 995-1006.	1.3	38
14	Split hand/foot malformation due to chromosome 7q aberrations(SHFM1): additional support for functional haploinsufficiency as the causative mechanism. European Journal of Human Genetics, 2009, 17, 1432-1438.	1.4	36
15	The inversa type of recessive dystrophic epidermolysis bullosa is caused by specific arginine and glycine substitutions in type VII collagen. Journal of Medical Genetics, 2011, 48, 160-167.	1.5	35
16	The molecular skin pathology of familial primary localized cutaneous amyloidosis. Experimental Dermatology, 2010, 19, 416-423.	1.4	34
17	Multigene panel sequencing of established and candidate melanoma susceptibility genes in a large cohort of Dutch nonâ€CDKN2A/CDK4 melanoma families. International Journal of Cancer, 2019, 144, 2453-2464.	2.3	33
18	<scp>RNA</scp>â€based therapies for genodermatoses. Experimental Dermatology, 2017, 26, 3-10.	1.4	28

#	Article	IF	Citations
19	Long-term follow-up of patients with recessive dystrophic epidermolysis bullosa in the Netherlands: Expansion of the mutation database and unusual phenotypeâ€“genotype correlations. Journal of Dermatological Science, 2009, 56, 9-18.	1.0	27
20	Natural Gene Therapy in Dystrophic Epidermolysis Bullosa. Archives of Dermatology, 2012, 148, 213.	1.7	27
21	Natural Exon Skipping Sets the Stage for Exon Skipping as Therapy for Dystrophic Epidermolysis Bullosa. Molecular Therapy - Nucleic Acids, 2019, 18, 465-475.	2.3	26
22	Cardiomyopathy in patients with epidermolysis bullosa simplex with mutations in KLHL24. British Journal of Dermatology, 2018, 179, 1181-1183.	1.4	23
23	A "late-but-fitter revertant cell" explains the high frequency of revertant mosaicism in epidermolysis bullosa. PLoS ONE, 2018, 13, e0192994.	1.1	18
24	Association of Epidermolysis Bullosa Simplex With Mottled Pigmentation andEXPH5Mutations. JAMA Dermatology, 2016, 152, 1137.	2.0	17
25	Generalized Ichthyotic Peeling Skin Syndrome due to FLG2 Mutations. Journal of Investigative Dermatology, 2018, 138, 1881-1884.	0.3	17
26	Hyperkeratotic hand eczema: Eczema or not?. Contact Dermatitis, 2020, 83, 196-205.	0.8	17
27	Somatic mosaicism for theCOL7A1mutation p.Gly2034Arg in the unaffected mother of a patient with dystrophic epidermolysis bullosa pruriginosa. British Journal of Dermatology, 2015, 172, 778-781.	1.4	14
28	Heterozygosity for a Novel Missense Mutation in theITGB4Gene Associated With Autosomal Dominant Epidermolysis Bullosa. JAMA Dermatology, 2016, 152, 558.	2.0	14
29	Epidermolysis Bullosa Simplex Caused by Distal Truncation of BPAG1-e: An Intermediate Generalized Phenotype with Prurigo Papules. Journal of Investigative Dermatology, 2017, 137, 2227-2230.	0.3	14
30	Genotype-phenotype correlations for pancreatic cancer risk in Dutch melanoma families with pathogenic CDKN2A variants. Journal of Medical Genetics, 2021, 58, 264-269.	1.5	13
31	Pancreatic cancer-associated gene polymorphisms in a nation-wide cohort of p16-Leiden germline mutation carriers; a caseâ€“control study. BMC Research Notes, 2015, 8, 264.	0.6	10
32	Murine type VII collagen distorts outcome in human skin graft mouse model for dystrophic epidermolysis bullosa. Experimental Dermatology, 2019, 28, 1153-1155.	1.4	7
33	A PLEC Isoform Identified in Skin, Muscle, and Heart. Journal of Investigative Dermatology, 2017, 137, 518-522.	0.3	6
34	Therapeutic Prospects of Exon Skipping for Epidermolysis Bullosa. International Journal of Molecular Sciences, 2021, 22, 12222.	1.8	6
35	Somatic mosaicism for the SALL1 mutation p.Ser371X in fullâ€blown Townesâ€“Brocks syndrome with Duane anomaly. American Journal of Medical Genetics, Part A, 2009, 149A, 812-815.	0.7	5
36	Diagnostic next generation sequencing in neonatal erythroderma. JDDG - Journal of the German Society of Dermatology, 2021, 19, 612-614.	0.4	5

#	Article	IF	Citations
37	Collodion babies: A 15-year retrospective multicenter study in The Netherlandsâ€"Evaluation of severity scores to predict the underlying disease. Journal of the American Academy of Dermatology, 2021, 84, 1111-1113.	0.6	4
38	The aggressive behaviour of squamous cell carcinoma in epidermolysis bullosa: analysis of clinical outcomes and tumour characteristics in the Dutch EB Registry. British Journal of Dermatology, 2022, 187, 824-826.	1.4	4
39	Design and Validation of a Conformation-Sensitive Capillary Electrophoresis System for Mutation Identification of the COL7A1 Gene with Automated Peak Comparison. Genetic Testing and Molecular Biomarkers, 2009, 13, 589-597.	0.3	3
40	In Vitro Models for the Evaluation of Antisense Oligonucleotides in Skin. Methods in Molecular Biology, 2022, 2434, 185-190.	0.4	2
41	Therapies for epidermolysis bullosa: delivery is key. British Journal of Dermatology, 2019, 180, 17-19.	1.4	1
42	Single glycine deletion in COL7A1 acting as glycine substitution in dystrophic epidermolysis bullosa. Journal of the European Academy of Dermatology and Venereology, 2021, 35, e597-e600.	1.3	1
43	In Vivo Models for the Evaluation of Antisense Oligonucleotides in Skin. Methods in Molecular Biology, 2022, 2434, 315-320.	0.4	1
44	Marcel F. Jonkman, MD, PhD (1957â€"2019). Journal of Investigative Dermatology, 2019, 139, 982-983.	0.3	0