Peter C Van Den Akker

List of Publications by Citations

43 papers	1,146 citations	19 h-index	33 g-index
46 ext. papers	1,412 ext. citations	4.3 avg, IF	3.76 L-index

#	Paper	IF	Citations
43	Loss-of-function variants in the filaggrin gene are a significant risk factor for peanut allergy. *Journal of Allergy and Clinical Immunology*, **2011**, 127, 661-7	11.5	342
42	WRN mutations in Werner syndrome patients: genomic rearrangements, unusual intronic mutations and ethnic-specific alterations. *Human Genetics*, **2010**, 128, 103-11	6.3	76
41	The international dystrophic epidermolysis bullosa patient registry: an online database of dystrophic epidermolysis bullosa patients and their COL7A1 mutations. *Human Mutation*, **2011**, 32, 1100-7	4.7	63
40	Improving the diagnostic yield of exome- sequencing by predicting gene-phenotype associations using large-scale gene expression analysis. *Nature Communications*, **2019**, 10, 2837	17.4	55
39	Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. *British Journal of Dermatology*, **2020**, 182, 574-592	4	45
38	Antisense Oligonucleotide-mediated Exon Skipping as a Systemic Therapeutic Approach for Recessive Dystrophic Epidermolysis Bullosa. *Molecular Therapy - Nucleic Acids*, **2016**, 5, e379	10.7	44
37	An overview and online registry of microvillus inclusion disease patients and their MYO5B mutations. *Human Mutation*, **2013**, 34, 1597-605	4.7	42
36	Left ventricular outflow tract obstruction: should cardiac screening be offered to first-degree relatives?. *Heart*, **2011**, 97, 1228-32	5.1	39
35	New ZMPSTE24 (FACE1) mutations in patients affected with restrictive dermopathy or related progeroid syndromes and mutation update. *European Journal of Human Genetics*, **2014**, 22, 1002-11	5.3	36
34	Mechanisms of natural gene therapy in dystrophic epidermolysis bullosa. *Journal of Investigative Dermatology*, **2014**, 134, 2097-2104	4.3	33
33	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, 1432-8	5.3	32
32	Analysis of the functional consequences of targeted exon deletion in COL7A1 reveals prospects for dystrophic epidermolysis bullosa therapy. *Molecular Therapy*, **2016**, 24, 1302-11	11.7	29
31	The molecular skin pathology of familial primary localized cutaneous amyloidosis. *Experimental Dermatology*, **2010**, 19, 416-23	4	28
30	-Related Cardiac Disease: Late Onset With a Variable and Mild Phenotype in a Large Cohort of Patients With the Lamin A/C p.(Arg331Gln) Founder Mutation. *Circulation: Cardiovascular Genetics*, **2017**, 10,		27
29	RNA-based therapies for genodermatoses. *Experimental Dermatology*, **2017**, 26, 3-10	4	25
28	Multigene panel sequencing of established and candidate melanoma susceptibility genes in a large cohort of Dutch non-CDKN2A/CDK4 melanoma families. *International Journal of Cancer*, **2019**, 144, 2453-2464	7.5	25
27	Natural gene therapy in dystrophic epidermolysis bullosa. *Archives of Dermatology*, **2012**, 148, 213-6		22

26	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, 160-7	5.8	21
25	Long-term follow-up of patients with recessive dystrophic epidermolysis bullosa in the Netherlands: expansion of the mutation database and unusual phenotype-genotype correlations. *Journal of Dermatological Science*, **2009**, 56, 9-18	4.3	20
24	Natural Exon Skipping Sets the Stage for Exon Skipping as Therapy for Dystrophic Epidermolysis Bullosa. *Molecular Therapy - Nucleic Acids*, **2019**, 18, 465-475	10.7	16
23	A "late-but-fitter revertant cell" explains the high frequency of revertant mosaicism in epidermolysis bullosa. *PLoS ONE*, **2018**, 13, e0192994	3.7	14
22	Association of Epidermolysis Bullosa Simplex With Mottled Pigmentation and EXPH5 Mutations. *JAMA Dermatology*, **2016**, 152, 1137-1141	5.1	14
21	Cardiomyopathy in patients with epidermolysis bullosa simplex with mutations in KLHL24. *British Journal of Dermatology*, **2018**, 179, 1181-1183	4	12
20	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	4.6	11
19	Generalized Ichthyotic Peeling Skin Syndrome due to FLG2 Mutations. *Journal of Investigative Dermatology*, **2018**, 138, 1881-1884	4.3	10
18	Heterozygosity for a Novel Missense Mutation in the ITGB4 Gene Associated With Autosomal Dominant Epidermolysis Bullosa. *JAMA Dermatology*, **2016**, 152, 558-62	5.1	10
17	Somatic mosaicism for the COL7A1 mutation p.Gly2034Arg in the unaffected mother of a patient with dystrophic epidermolysis bullosa pruriginosa. *British Journal of Dermatology*, **2015**, 172, 778-81	4	9
16	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	4.3	8
15	Pancreatic cancer-associated gene polymorphisms in a nation-wide cohort of p16-Leiden germline mutation carriers; a case-control study. *BMC Research Notes*, **2015**, 8, 264	2.3	7
14	Murine type VII collagen distorts outcome in human skin graft mouse model for dystrophic epidermolysis bullosa. *Experimental Dermatology*, **2019**, 28, 1153-1155	4	6
13	Hyperkeratotic hand eczema: Eczema or not?. *Contact Dermatitis*, **2020**, 83, 196-205	2.7	5
12	Somatic mosaicism for the SALL1 mutation p.Ser371X in full-blown Townes-Brocks syndrome with Duane anomaly. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 812-5	2.5	5
11	Improving the diagnostic yield of exome-sequencing, by predicting gene-phenotype associations using large-scale gene expression analysis		5
10	Genotype-phenotype correlations for pancreatic cancer risk in Dutch melanoma families with pathogenic variants. *Journal of Medical Genetics*, **2021**, 58, 264-269	5.8	4
9	A PLEC Isoform Identified in Skin, Muscle, and Heart. *Journal of Investigative Dermatology*, **2017**, 137, 518-522	4.3	3

8	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, 589-97	1.6	1
7	Collodion babies: A 15-year retrospective multicenter study in The Netherlands-Evaluation of severity scores to predict the underlying disease. *Journal of the American Academy of Dermatology*, **2021**, 84, 1111-1113	4.5	1
6	Diagnostic next generation sequencing in neonatal erythroderma. *JDDG - Journal of the German Society of Dermatology*, **2021**, 19, 612-614	1.2	1
5	In Vitro Models for the Evaluation of Antisense Oligonucleotides in Skin.. *Methods in Molecular Biology*, **2022**, 2434, 185-190	1.4	0
4	Marcel F. Jonkman, MD, PhD (1957–2019). *Journal of Investigative Dermatology*, **2019**, 139, 982-983	4.3	
3	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	4.6	
2	Therapies for epidermolysis bullosa: delivery is key. *British Journal of Dermatology*, **2019**, 180, 17-19	4	
1	In Vivo Models for the Evaluation of Antisense Oligonucleotides in Skin.. *Methods in Molecular Biology*, **2022**, 2434, 315-320	1.4	