

Hassan Vahidnezhad

List of Publications by Year in descending order

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Version: 2024-02-01

105
papers

1,667
citations

331670

21
h-index

361022

35
g-index

105
all docs

105
docs citations

105
times ranked

2099
citing authors

#	ARTICLE	IF	CITATIONS
1	Klippel-ÉTrenaunay syndrome belongs to the <i>PIK3CA</i>-related overgrowth spectrum (PROS). <i>Experimental Dermatology</i> , 2016, 25, 17-19.	2.9	143
2	The human CIB1-ÉEVER1-ÉEVER2 complex governs keratinocyte-intrinsic immunity to É2-papillomaviruses. <i>Journal of Experimental Medicine</i> , 2018, 215, 2289-2310.	8.5	92
3	Molecular pathology of the basement membrane zone in heritable blistering diseases. <i>Matrix Biology</i> , 2017, 57-58, 76-85.	3.6	58
4	Applications of Spherical Nucleic Acid Nanoparticles as Delivery Systems. <i>Trends in Molecular Medicine</i> , 2019, 25, 1066-1079.	6.7	58
5	Humans with inherited T-Cell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. <i>Cell</i> , 2021, 184, 3812-3828.e30.	28.9	53
6	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.	3.6	49
7	Genotype-Éphenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. <i>British Journal of Dermatology</i> , 2020, 182, 729-737.	1.5	47
8	Research Techniques Made Simple: Whole-Transcriptome Sequencing by RNA-Seq for ÉDiagnosis of Monogenic Disorders. <i>Journal of Investigative Dermatology</i> , 2020, 140, 1117-1126.e1.	0.7	46
9	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.	3.6	45
10	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.	0.7	44
11	Autosomal recessive congenital ichthyosis: Genomic landscape and phenotypic spectrum in a cohort of 125 consanguineous families. <i>Human Mutation</i> , 2019, 40, 288-298.	2.5	43
12	Skin Manifestations in COVID-19 Patients: Are They Indicators for Disease Severity? A Systematic Review. <i>Frontiers in Medicine</i> , 2021, 8, 634208.	2.6	42
13	Inherited non-alcoholic fatty liver disease and dyslipidemia due to monoallelic ABHD5 mutations. <i>Journal of Hepatology</i> , 2019, 71, 366-370.	3.7	41
14	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.	0.7	39
15	Research Techniques Made Simple: Genome-Wide Homozygosity/Autozygosity Mapping Is a Powerful Tool Éfor Identifying Candidate Genes in Autosomal Recessive Genetic Diseases. <i>Journal of Investigative Dermatology</i> , 2018, 138, 1893-1900.	0.7	36
16	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.	0.7	35
17	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.	0.7	31
18	Next generation sequencing identifies double homozygous mutations in two distinct genes (<i>EXPH5</i> and <i>COL17A1</i>) in a patient with concomitant simplex and junctional epidermolysis bullosa. <i>Human Mutation</i> , 2018, 39, 1349-1354.	2.5	29

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19	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.	0.7	28
20	The Kindler Syndrome: A Spectrum of FERMT1 Mutations in Iranian Families. <i>Journal of Investigative Dermatology</i> , 2015, 135, 1447-1450.	0.7	26
21	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.	5.8	22
22	Lipoid proteinosis: phenotypic heterogeneity in Iranian families with c.507delT mutation in <i>ECM1</i> . <i>Experimental Dermatology</i> , 2015, 24, 220-222.	2.9	20
23	Fibroadipose Hyperplasia versus Proteus Syndrome: Segmental Overgrowth with a Mosaic Mutation in the PIK3CA Gene. <i>Journal of Investigative Dermatology</i> , 2015, 135, 1450-1453.	0.7	20
24	Pachyonychia congenita: a case report of a successful treatment with rosuvastatin in a patient with a <i>KRT6A</i> mutation. <i>British Journal of Dermatology</i> , 2019, 181, 584-586.	1.5	20
25	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.	0.7	19
26	Phenotypic spectrum of autosomal recessive congenital ichthyosis due to <i>PNPLA1</i> mutation. <i>British Journal of Dermatology</i> , 2017, 177, 319-322.	1.5	19
27	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.	2.8	19
28	A CIB1 Splice-Site Founder Mutation in Families with Atypical Epidermodysplasia Verruciformis. <i>Journal of Investigative Dermatology</i> , 2019, 139, 1195-1198.	0.7	19
29	Epidermodysplasia Verruciformis: Genetic Heterogeneity and EVER1 and EVER2 Mutations Revealed by Genome-Wide Analysis. <i>Journal of Investigative Dermatology</i> , 2019, 139, 241-244.	0.7	19
30	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.	2.9	19
31	Whole Genome Linkage Analysis Followed by Whole Exome Sequencing Identifies Nicastrin (NCSTN) as a Causative Gene in a Multiplex Family with I ³ -Secretase Spectrum of Autoinflammatory Skin Phenotypes. <i>Journal of Investigative Dermatology</i> , 2016, 136, 1283-1286.	0.7	17
32	Expanding the Genotypic Spectrum of Bathing Suit Ichthyosis. <i>JAMA Dermatology</i> , 2017, 153, 537.	4.1	17
33	Molecular Genetics of Keratinization Disorders – What’s New About Ichthyosis. <i>Acta Dermato-Venereologica</i> , 2020, 100, adv00095-185.	1.3	17
34	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.	2.7	16
35	A novel autosomal recessive <i>GJB2</i> -associated disorder: Ichthyosis follicularis, bilateral severe sensorineural hearing loss, and punctate palmoplantar keratoderma. <i>Human Mutation</i> , 2019, 40, 217-229.	2.5	16
36	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.	3.2	16

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37	Biallelic KRT5 mutations in autosomal recessive epidermolysis bullosa simplex, including a complete human keratin 5 knock-out. Matrix Biology, 2019, 83, 48-59.	3.6	15
38	Gene-Targeted Next-Generation Sequencing Identifies a Novel CLDN1 Mutation in a Consanguineous Family With NISCH Syndrome. American Journal of Gastroenterology, 2017, 112, 396-398.	0.4	14
39	Molecular genetics of a cohort of 635 cases of phenylketonuria in a consanguineous population. Journal of Inherited Metabolic Disease, 2018, 41, 1159-1167.	3.6	14
40	ENPP1 variants in patients with GACI and PXE expand the clinical and genetic heterogeneity of heritable disorders of ectopic calcification. PLoS Genetics, 2022, 18, e1010192.	3.5	13
41	Genotypic Heterogeneity and the Mode of Inheritance in Epidermolysis Bullosa. JAMA Dermatology, 2016, 152, 517.	4.1	11
42	Genomics-based treatment in a patient with two overlapping heritable skin disorders: Epidermolysis bullosa and acrodermatitis enteropathica. Human Mutation, 2020, 41, 906-912.	2.5	11
43	Using immunofluorescence (antigen) mapping in the diagnosis and classification of epidermolysis bullosa: a first report from Iran. International Journal of Dermatology, 2015, 54, e416-23.	1.0	10
44	Phenotypic heterogeneity in PIK3CA-related overgrowth spectrum. British Journal of Dermatology, 2016, 175, 810-814.	1.5	10
45	Mycophenolate mofetil treatment of an H syndrome patient with a SLC29A3 mutation. Dermatologic Therapy, 2020, 33, e14375.	1.7	10
46	Association of MTHFR C677T polymorphism with elevated homocysteine level and disease development in vitiligo. International Journal of Immunogenetics, 2020, 47, 342-350.	1.8	10
47	Homozygous MEFV Gene Variant and Pyrin-Associated Autoinflammation With Neutrophilic Dermatitis. JAMA Dermatology, 2021, 157, 1466.	4.1	10
48	Recalcitrant Warts, Epidermodysplasia Verruciformis, and the Tree-Man Syndrome: Phenotypic Spectrum of Cutaneous Human Papillomavirus Infections at the Intersection of Genetic Variability of Viral and Human Genomes. Journal of Investigative Dermatology, 2022, 142, 1265-1269.	0.7	10
49	Losartan treatment improves recessive dystrophic epidermolysis bullosa: A case series. Dermatologic Therapy, 2022, 35, e15515.	1.7	10
50	Hyaline Fibromatosis Syndrome: A Novel Mutation and Recurrent Founder Mutation in the CMG2/ANTXR2 Gene. Acta Dermato-Venereologica, 2017, 97, 108-109.	1.3	9
51	Homozygous IL1RN Mutation in Siblings with Deficiency of Interleukin-1 Receptor Antagonist (DIRA). Journal of Clinical Immunology, 2020, 40, 637-642.	3.8	9
52	Interpretation of genomic sequence variants in heritable skin diseases: A primer for clinicians. Journal of the American Academy of Dermatology, 2023, 89, 569-576.	1.2	9
53	A novel mutation in ST14 at a functionally significant amino acid residue expands the spectrum of ichthyosis-hypotrichosis syndrome. Orphanet Journal of Rare Diseases, 2017, 12, 176.	2.7	8
54	The utility of dermal fibroblasts in treatment of skin disorders: A paradigm of recessive dystrophic epidermolysis bullosa. Dermatologic Therapy, 2021, 34, e15028.	1.7	8

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55	Infantile systemic hyalinosis in an Iranian family with a mutation in the <i>CMG2/ANTXR2</i> gene. <i>Clinical and Experimental Dermatology</i> , 2015, 40, 636-639.	1.3	7
56	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.	0.9	7
57	Erythrokeratoderma: a manifestation associated with multiple types of ichthyoses with different gene defects. <i>British Journal of Dermatology</i> , 2018, 178, e219-e221.	1.5	7
58	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	7
59	Hypotrichosis with juvenile macular dystrophy: Combination of whole-genome sequencing and genome-wide homozygosity mapping identifies a large deletion in <i>CDH3</i> initially undetected by whole-exome sequencing. A lesson from next-generation sequencing. <i>Molecular Genetics & Genomic Medicine</i> . 2019, 7, e975.	1.2	7
60	Arrhythmogenic right ventricular cardiomyopathy in patients with biallelic JUP-associated skin fragility. <i>Scientific Reports</i> , 2020, 10, 21622.	3.3	7
61	Homozygous ITGA3 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.	0.7	7
62	Pathogenic <i>DST</i> sequence variants result in either epidermolysis bullosa simplex (EBS) or hereditary sensory and autonomic neuropathy type 6 (HSAN6). <i>Experimental Dermatology</i> , 2022, 31, 949-955.	2.9	7
63	Evaluation of neurodevelopmental symptoms in 10 cases of neonatal ichthyosis and sclerosing cholangitis syndrome. <i>Pediatric Dermatology</i> , 2022, 39, 590-593.	0.9	7
64	De novo severe pemphigus vulgaris following SARS-CoV-2 vaccination with BBIBP-CorV. <i>Dermatologic Therapy</i> , 2022, , e15448.	1.7	7
65	Seven novel <i>COL7A1</i> mutations identified in patients with recessive dystrophic epidermolysis bullosa from Mexico. <i>Clinical and Experimental Dermatology</i> , 2018, 43, 579-584.	1.3	6
66	Increased level of cathelicidin (LL37) in vitiligo: Possible pathway independent from vitamin D receptor gene polymorphism. <i>Experimental Dermatology</i> , 2020, 29, 1176-1185.	2.9	6
67	The quality of life in epidermolysis bullosa (EB-QoL) questionnaire: Translation, cultural adaptation, and validation into the Farsi language. <i>International Journal of Women's Dermatology</i> , 2020, 6, 301-305.	2.0	6
68	The matriptase-prostasin proteolytic cascade in dermatologic diseases. <i>Experimental Dermatology</i> , 2020, 29, 580-587.	2.9	6
69	BMI1 and TWIST1 Downregulated mRNA Expression in Basal Cell Carcinoma. <i>Asian Pacific Journal of Cancer Prevention</i> , 2014, 15, 3797-3800.	1.2	6
70	Whole-transcriptome sequencing-based concomitant detection of viral and human genetic determinants of cutaneous lesions. <i>JCI Insight</i> , 2022, 7, .	5.0	6
71	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.	0.7	5
72	Widespread aplasia cutis congenita in sibs with <i>PLEC1</i> and <i>ITGB4</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1547-1555.	1.2	5

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73	Keratitis&chthyosis&deafness syndrome: Phenotypic heterogeneity and treatment perspective of patients with p. <sc> Asp50Asn <i>GJB2</i> </sc> mutation. <i>Dermatologic Therapy</i> , 2020, 33, e14493.	1.7	5
74	Losartan for treatment of epidermolysis bullosa: A new perspective. <i>Dermatologic Therapy</i> , 2021, 34, e14638.	1.7	5
75	Self-Reported Hand Eczema: Assessment of Prevalence and Risk Factors in Health Care Versus Non&Health Care Workers During the COVID-19 Pandemic. <i>Dermatitis</i> , 2021, 32, e19-e21.	1.6	5
76	Genetic heterogeneity of heritable ectopic mineralization disorders in a large international cohort. <i>Genetics in Medicine</i> , 2022, 24, 75-86.	2.4	5
77	Pathomechanisms of epidermolysis bullosa: Beyond structural proteins. <i>Matrix Biology</i> , 2022, 110, 91-105.	3.6	5
78	Expanding genetics and phenotypic spectrum of epidermodysplasia verruciformis. <i>British Journal of Dermatology</i> , 2016, 175, 1138-1139.	1.5	4
79	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.	0.8	4
80	Coronavirus disease 2019 and epidermolysis bullosa: Report of three cases. <i>Dermatologic Therapy</i> , 2020, 33, e14194.	1.7	4
81	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> , 2021, 99, 43-57.	3.6	4
82	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.	0.3	4
83	Knockdown of SDR9C7 Impairs Epidermal Barrier Function. <i>Journal of Investigative Dermatology</i> , 2021, 141, 1754-1764.e1.	0.7	4
84	Recalcitrant Cutaneous Warts in a Family with Inherited ICOS Deficiency. <i>Journal of Investigative Dermatology</i> , 2022, 142, 2435-2445.	0.7	4
85	CD147 inhibitors as a treatment for melanoma: Promising agents against SARS&CoV infection. <i>Dermatologic Therapy</i> , 2020, 33, e14449.	1.7	3
86	Epidermolysis bullosa and the COVID-19 pandemic: challenges and recommendations. <i>Journal of Dermatological Treatment</i> , 2020, , 1-2.	2.2	3
87	Modeling breast acini in tissue culture for detection of malignant phenotype reversion to non-malignant phenotype. <i>Iranian Biomedical Journal</i> , 2009, 13, 191-8.	0.7	3
88	Whole&transcriptome sequencing identifies postzygotic <i>ATP2A2</i> mutations in a patient misdiagnosed with herpes zoster, confirming the diagnosis of very late&onset segmental Darier disease. <i>Experimental Dermatology</i> , 2022, 31, 943-948.	2.9	3
89	Ichthyosis, psoriasiform dermatitis, and recurrent fungal infections in patients with biallelic mutations in <i>PERP</i>. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2022, 36, 472-479.	2.4	3
90	Autozygosity Mapping by Genome-wide Single Nucleotide Polymorphism Array Identifies a Novel Homozygous HR Mutation in a Consanguineous Family with Universal Hereditary Hair Loss. <i>International Journal of Dermatology and Venereology</i> , 2021, 4, 82-85.	0.3	2

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91	Very-Early-Onset Inflammatory Bowel Disease in a Patient With Junctional Epidermolysis Bullosa With a Homozygous Mutation in the $\beta 6$ Integrin Gene (<i>ITGA6</i>). <i>Inflammatory Bowel Diseases</i> , 2021, 27, 1865-1869.	1.9	2
92	Novel splice mutation in CDSN gene causing type b peeling skin syndrome. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2022, 36, .	2.4	2
93	Ichthyosis follicularis syndromes in patients with germline mutations in <i>GJB2</i> . <i>Clinical and Experimental Dermatology</i> , 2022, , .	1.3	2
94	Expanding mutation landscape and phenotypic spectrum of autosomal recessive congenital ichthyosis. <i>British Journal of Dermatology</i> , 2017, 177, 342-343.	1.5	1
95	639 Homozygous mutation in ITK associated with monogenic inborn errors of immunity underlies susceptibility to human papilloma virus infections (epidermodysplasia verruciformis). <i>Journal of Investigative Dermatology</i> , 2017, 137, S110.	0.7	1
96	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.	1.3	1
97	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.	1.2	1
98	509 Disease-targeted next generation sequencing identifies mutations in patients with epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , 2017, 137, S87.	0.7	0
99	793 Genome-wide single nucleotide polymorphism-based autozygosity mapping facilitates identification of mutations in consanguineous families with epidermolysis bullosa. <i>Journal of Investigative Dermatology</i> , 2018, 138, S135.	0.7	0
100	A study of gene mutations and how they relate to the different types of ichthyosis. <i>British Journal of Dermatology</i> , 2020, 182, e101.	1.5	0
101	Management of symptomatic mucosal involvement in paediatric pachyonychia congenita. <i>British Journal of Dermatology</i> , 2020, 182, 536-537.	1.5	0
102	Lipoid Proteinosis Due to Homozygous Deletion Mutation (c.735delTG) in the ECM1 Gene Presents with Seizures and Hoarseness but No Skin Involvement. <i>International Journal of Dermatology and Venereology</i> , 2020, 3, 43-45.	0.3	0
103	Linear basal cell nevus with a novel mosaic <i>PTCH1</i> mutation. <i>Experimental Dermatology</i> , 2020, 29, 531-534.	2.9	0
104	186 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>Journal of Investigative Dermatology</i> , 2021, 141, S33.	0.7	0
105	Are Dyskeratosis Congenita patients at higher risk of symptomatic COVID-19?. <i>Medical Hypotheses</i> , 2022, 163, 110843.	1.5	0