Hassan Vahidnezhad

List of Publications by Year
in descending order

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	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
2	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
3	Genetic heterogeneity of heritable ectopic mineralization disorders in a large international cohort. Genetics in Medicine, 2022, 24, 75-86.	2.4	5
4	Novel splice mutation in CDSN gene causing type b peeling skin syndrome. Journal of the European Academy of Dermatology and Venereology, 2022, 36, .	2.4	2
5	Pathogenic DST sequence variants result in either epidermolysis bullosa simplex (EBS) or hereditary sensory and autonomic neuropathy type 6 (HSANâ€VI). Experimental Dermatology, 2022, 31, 949-955.	2.9	7
6	Whole-transcriptome sequencingâ€“based concomitant detection of viral and human genetic determinants of cutaneous lesions. JCI Insight, 2022, 7, .	5.0	6
7	Recalcitrant Cutaneous Warts in a Family with Inherited ICOS Deficiency. Journal of Investigative Dermatology, 2022, 142, 2435-2445.	0.7	4
8	Evaluation of neurodevelopmental symptoms in 10 cases of neonatal ichthyosis and sclerosing cholangitis syndrome. Pediatric Dermatology, 2022, 39, 590-593.	0.9	7
9	Wholeâ€transcriptome sequencing identifies postzygotic ATP2A2 mutations in a patient misdiagnosed with herpes zoster, confirming the diagnosis of very lateâ€onset segmental Darier disease. Experimental Dermatology, 2022, 31, 943-948.	2.9	3
10	De novo severe pemphigus vulgaris following <scp>SARSâ€CoV</scp> â€2 vaccination with <scp>BBIBPâ€CorV</scp>. Dermatologic Therapy, 2022, , e15448.	1.7	7
11	Ichthyosis follicularis syndromes in patients with germline mutations in <scp> GJB2 </scp>. Clinical and Experimental Dermatology, 2022, , .	1.3	2
12	Ichthyosis, psoriasiform dermatitis, and recurrent fungal infections in patients with biallelic mutations in PERP. Journal of the European Academy of Dermatology and Venereology, 2022, 36, 472-479.	2.4	3
13	Losartan treatment improves recessive dystrophic epidermolysis bullosa: A case series. Dermatologic Therapy, 2022, 35, e15515.	1.7	10
14	Are Dyskeratosis Congenita patients at higher risk of symptomatic COVID-19?. Medical Hypotheses, 2022, 163, 110843.	1.5	0
15	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
16	Pathomechanisms of epidermolysis bullosa: Beyond structural proteins. Matrix Biology, 2022, 110, 91-105.	3.6	5
17	Losartan for treatment of epidermolysis bullosa: A new perspective. Dermatologic Therapy, 2021, 34, e14638.	1.7	5
18	Skin Manifestations in COVID-19 Patients: Are They Indicators for Disease Severity? A Systematic Review. Frontiers in Medicine, 2021, 8, 634208.	2.6	42

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19	186 Kindler epidermolysis bullosa-like skin phenotype and downregulated basement membrane zone gene expression in poikiloderma with neutropenia and a homozygous USB1 mutation. Journal of Investigative Dermatology, 2021, 141, S33.	0.7	0
20	Autozygosity Mapping by Genome-wide Single Nucleotide Polymorphism Array Identifies a Novel Homozygous HR Mutation in a Consanguineous Family with Universal Hereditary Hair Loss. International Journal of Dermatology and Venereology, 2021, 4, 82-85.	0.3	2
21	Kindler epidermolysis bullosa-like skin phenotype and downregulated basement membrane zone gene expression in poikiloderma with neutropenia and a homozygous USB1 mutation. Matrix Biology, 2021, 99, 43-57.	3.6	4
22	Homozygous ITGA3 Missense Mutation in Adults in a Family with Syndromic Epidermolysis Bullosa (ILNEB) without Pulmonary Involvement. Journal of Investigative Dermatology, 2021, 141, 2752-2756.	0.7	7
23	Whole-Transcriptome Analysis by RNA Sequencing for Genetic Diagnosis of Mendelian Skin Disorders in the Context of Consanguinity. Clinical Chemistry, 2021, 67, 876-888.	3.2	16
24	Genetic Predisposition to Numerous Large Ulcerating Basal Cell Carcinomas and Response to Immune Therapy. International Journal of Dermatology and Venereology, 2021, 4, 70-75.	0.3	4
25	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
26	Knockdown of SDR9C7 Impairs Epidermal Barrier Function. Journal of Investigative Dermatology, 2021, 141, 1754-1764.e1.	0.7	4
27	Humans with inherited TÂcell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. Cell, 2021, 184, 3812-3828.e30.	28.9	53
28	Ancestral patterns of recessive dystrophic epidermolysis bullosa mutations in Hispanic populations suggest sephardic ancestry. American Journal of Medical Genetics, Part A, 2021, 185, 3390-3400.	1.2	1
29	Very-Early-Onset Inflammatory Bowel Disease in a Patient With Junctional Epidermolysis Bullosa With a Homozygous Mutation in the Î±6 Integrin Gene (ITGA6). Inflammatory Bowel Diseases, 2021, 27, 1865-1869.	1.9	2
30	Homozygous MEFV Gene Variant and Pyrin-Associated Autoinflammation With Neutrophilic Dermatosis. JAMA Dermatology, 2021, 157, 1466.	4.1	10
31	Self-Reported Hand Eczema: Assessment of Prevalence and Risk Factors in Health Care Versus Nonâ€“Health Care Workers During the COVID-19 Pandemic. Dermatitis, 2021, 32, e19-e21.	1.6	5
32	Genotypeâ€“phenotype correlation in a large English cohort of patients with autosomal recessive ichthyosis. British Journal of Dermatology, 2020, 182, 729-737.	1.5	47
33	Increased level of cathelicidin (LLâ€37) in vitiligo: Possible pathway independent from vitamin D receptor gene polymorphism. Experimental Dermatology, 2020, 29, 1176-1185.	2.9	6
34	Coronavirus disease 2019 and epidermolysis bullosa: Report of three cases. Dermatologic Therapy, 2020, 33, e14194.	1.7	4
35	Keratitisâ€ichthyosisâ€deafness syndrome: Phenotypic heterogeneity and treatment perspective of patients with p. <scp> Asp50Asn GJB2 </scp> mutation. Dermatologic Therapy, 2020, 33, e14493.	1.7	5
36	Mycophenolate mofetil treatment of an H syndrome patient with a <scp> SLC29A3 </scp> mutation. Dermatologic Therapy, 2020, 33, e14375.	1.7	10

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37	CD147 inhibitors as a treatment for melanoma: Promising agents against SARSâ€CoV â€2 infection. Dermatologic Therapy, 2020, 33, e14449.	1.7	3
38	Arrhythmogenic right ventricular cardiomyopathy in patients with biallelic JUP-associated skin fragility. Scientific Reports, 2020, 10, 21622.	3.3	7
39	Research Techniques Made Simple: Whole-Transcriptome Sequencing by RNA-Seq forÂDiagnosis of Monogenic Disorders. Journal of Investigative Dermatology, 2020, 140, 1117-1126.e1.	0.7	46
40	The quality of life in epidermolysis bullosa (EB-QoL) questionnaire: Translation, cultural adaptation, and validation into the Farsi language. International Journal of Women's Dermatology, 2020, 6, 301-305.	2.0	6
41	A study of gene mutations and how they relate to the different types of ichthyosis. British Journal of Dermatology, 2020, 182, e101.	1.5	0
42	Homozygous IL1RN Mutation in Siblings with Deficiency of Interleukin-1 Receptor Antagonist (DIRA). Journal of Clinical Immunology, 2020, 40, 637-642.	3.8	9
43	Epidermolysis bullosa and the COVID-19 pandemic: challenges and recommendations. Journal of Dermatological Treatment, 2020, , 1-2.	2.2	3
44	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
45	Management of symptomatic mucosal involvement in paediatric pachyonychia congenita. British Journal of Dermatology, 2020, 182, 536-537.	1.5	0
46	Genomicsâ€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
47	Lipoid Proteinosis Due to Homozygous Deletion Mutation (c.735delTG) in the ECM1 Gene Presents with Seizures and Hoarseness but No Skin Involvement. International Journal of Dermatology and Venereology, 2020, 3, 43-45.	0.3	0
48	The matriptaseâ€prostasin proteolytic cascade in dermatologic diseases. Experimental Dermatology, 2020, 29, 580-587.	2.9	6
49	Linear basal cell nevus with a novel mosaic PTCH1 mutation. Experimental Dermatology, 2020, 29, 531-534.	2.9	0
50	Molecular Genetics of Keratinization Disorders â€“ Whatâ€™s New About Ichthyosis. Acta Dermato-Venereologica, 2020, 100, adv00095-185.	1.3	17
51	Biallelic KRT5 mutations in autosomal recessive epidermolysis bullosa simplex, including a complete human keratin 5 â€œknock-outâ€. Matrix Biology, 2019, 83, 48-59.	3.6	15
52	Assessment of the risk and characterization of non-melanoma skin cancer in Kindler syndrome: study of a series of 91 patients. Orphanet Journal of Rare Diseases, 2019, 14, 183.	2.7	16
53	Hypotrichosis with juvenile macular dystrophy: Combination of wholeâ€genome sequencing and genomeâ€wide homozygosity mapping identifies a large deletion in CDH3 initially undetected by wholeâ€exome sequencingâ€”A lesson from nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2019, 7, e975.	1.2	7
54	Applications of Spherical Nucleic Acid Nanoparticles as Delivery Systems. Trends in Molecular Medicine, 2019, 25, 1066-1079.	6.7	58

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55	Widespread aplasia cutis congenita in sibs with PLEC1 and ITGB4 variants. American Journal of Medical Genetics, Part A, 2019, 179, 1547-1555.	1.2	5
56	Inherited non-alcoholic fatty liver disease and dyslipidemia due to monoallelic ABHD5 mutations. Journal of Hepatology, 2019, 71, 366-370.	3.7	41
57	A CIB1 Splice-Site Founder Mutation in Families withÂTypical Epidermodysplasia Verruciformis. Journal of Investigative Dermatology, 2019, 139, 1195-1198.	0.7	19
58	Inherited Interleukin 2â€“Inducible T-Cell (ITK) Kinase Deficiency in Siblings With Epidermodysplasia Verruciformis and Hodgkin Lymphoma. Clinical Infectious Diseases, 2019, 68, 1938-1941.	5.8	22
59	Phenotypic Spectrum of Epidermolysis Bullosa: The Paradigm of Syndromic versus Non-Syndromic Skin Fragility Disorders. Journal of Investigative Dermatology, 2019, 139, 522-527.	0.7	39
60	Autosomal recessive congenital ichthyosis: Genomic landscape and phenotypic spectrum in a cohort of 125 consanguineous families. Human Mutation, 2019, 40, 288-298.	2.5	43
61	A novel autosomal recessiveGJB2-associated disorder: Ichthyosis follicularis, bilateral severe sensorineural hearing loss, and punctate palmoplantar keratoderma. Human Mutation, 2019, 40, 217-229.	2.5	16
62	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. Matrix Biology, 2019, 81, 91-106.	3.6	45
63	Epidermodysplasia Verruciformis: Genetic Heterogeneity and EVER1 and EVER2 Mutations Revealed by Genome-Wide Analysis. Journal of Investigative Dermatology, 2019, 139, 241-244.	0.7	19
64	Pachyonychia congenita: a case report of a successful treatment with rosuvastatin in a patient with a KRT6A mutation. British Journal of Dermatology, 2019, 181, 584-586.	1.5	20
65	Genomeâ€wide single nucleotide polymorphismâ€based autozygosity mapping facilitates identification of mutations in consanguineous families with epidermolysis bullosa. Experimental Dermatology, 2019, 28, 1118-1121.	2.9	19
66	Seven novel COL7A1 mutations identified in patients with recessive dystrophic epidermolysis bullosa from Mexico. Clinical and Experimental Dermatology, 2018, 43, 579-584.	1.3	6
67	First report of COL7A1 mutations in two patients with recessive dystrophic epidermolysis bullosa from Peru. Clinical and Experimental Dermatology, 2018, 43, 719-722.	1.3	1
68	Erythrokeratoderma: a manifestation associated with multiple types of ichthyoses with different gene defects. British Journal of Dermatology, 2018, 178, e219-e221.	1.5	7
69	Recessive mutation in tetraspanin CD151 causes Kindler syndrome-like epidermolysis bullosa with multi-systemic manifestations including nephropathy. Matrix Biology, 2018, 66, 22-33.	3.6	49
70	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
71	The human CIB1â€“EVER1â€“EVER2 complex governs keratinocyte-intrinsic immunity to Î²-papillomaviruses. Journal of Experimental Medicine, 2018, 215, 2289-2310.	8.5	92
72	The genetic basis of hyaline fibromatosis syndrome in patients from a consanguineous background: a case series. BMC Medical Genetics, 2018, 19, 87.	2.1	7

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73	Next generation sequencing identifies double homozygous mutations in two distinct genes (EXPH5 and COL17A1) in a patient with concomitant simplex and junctional epidermolysis bullosa. Human Mutation, 2018, 39, 1349-1354.	2.5	29
74	Research Techniques Made Simple: Genome-Wide Homozygosity/Autozygosity Mapping Is a Powerful ToolÂfor Identifying Candidate Genes in Autosomal Recessive Genetic Diseases. Journal of Investigative Dermatology, 2018, 138, 1893-1900.	0.7	36
75	793 Genome-wide single nucleotide polymorphism-based autozygosity mapping facilitates identification of mutations in consanguineous families with epidermolysis bullosa. Journal of Investigative Dermatology, 2018, 138, S135.	0.7	0
76	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
77	Expanding the Genotypic Spectrum of Bathing Suit Ichthyosis. JAMA Dermatology, 2017, 153, 537.	4.1	17
78	Phenotypic spectrum of autosomal recessive congenital ichthyosis due to PNPLA1 mutation. British Journal of Dermatology, 2017, 177, 319-322.	1.5	19
79	Multigene Next-Generation Sequencing Panel Identifies Pathogenic Variants in Patients with Unknown Subtype of Epidermolysis Bullosa: Subclassification with Prognostic Implications. Journal of Investigative Dermatology, 2017, 137, 2649-2652.	0.7	31
80	Expanding mutation landscape and phenotypic spectrum of autosomal recessive congenital ichthyosis. British Journal of Dermatology, 2017, 177, 342-343.	1.5	1
81	Autosomal recessive congenital ichthyosis: CERS3 mutations identified by a next generation sequencing panel targeting ichthyosis genes. European Journal of Human Genetics, 2017, 25, 1282-1285.	2.8	19
82	509 Disease-targeted next generation sequencing identifies mutations in patients with epidermolysis bullosa. Journal of Investigative Dermatology, 2017, 137, S87.	0.7	0
83	639 Homozygous mutation in ITK associated with monogenic inborn errors of immunity underlies susceptibility to human papilloma virus infections (epidermodysplasia verruciformis). Journal of Investigative Dermatology, 2017, 137, S110.	0.7	1
84	Molecular Dynamics Simulation of the Consequences of a PYCR1 Mutation (p.Ala189Val) in Patients with Complex Connective Tissue Disorder and Severe Intellectual Disability. Journal of Investigative Dermatology, 2017, 137, 525-528.	0.7	5
85	Dystrophic Epidermolysis Bullosa: COL7A1 Mutation Landscape in a Multi-Ethnic Cohort of 152 Extended Families with High Degree of Customary Consanguineous Marriages. Journal of Investigative Dermatology, 2017, 137, 660-669.	0.7	44
86	Molecular pathology of the basement membrane zone in heritable blistering diseases:. Matrix Biology, 2017, 57-58, 76-85.	3.6	58
87	Gene-Targeted Next Generation Sequencing Identifies PNPLA1 Mutations in Patients with a Phenotypic Spectrum of Autosomal Recessive Congenital Ichthyosis: The Impact of Consanguinity. Journal of Investigative Dermatology, 2017, 137, 678-685.	0.7	28
88	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
89	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
90	Phenotypic heterogeneity inPIK3CA-related overgrowth spectrum. British Journal of Dermatology, 2016, 175, 810-814.	1.5	10

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91	Expanding genetics and phenotypic spectrum of epidermodysplasia verruciformis. British Journal of Dermatology, 2016, 175, 1138-1139.	1.5	4
92	Whole Genome Linkage Analysis Followed by Whole Exome Sequencing Identifies Nicastrin (NCSTN) as a Causative Gene in a Multiplex Family with Î³-Secretase Spectrum of Autoinflammatory Skin Phenotypes. Journal of Investigative Dermatology, 2016, 136, 1283-1286.	0.7	17
93	Co-existence of phenylketonuria either with maple syrup urine disease or Sandhoff disease in two patients from Iran: emphasizing the role of consanguinity. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 1215-1219.	0.9	7
94	Klippelâ€“Trenaunay syndrome belongs to the PIK3CAâ€related overgrowth spectrum (PROS). Experimental Dermatology, 2016, 25, 17-19.	2.9	143
95	Kindler syndrome, an orphan disease of cell/matrix adhesion in the skin â€“ molecular genetics and therapeutic opportunities. Expert Opinion on Orphan Drugs, 2016, 4, 845-854.	0.8	4
96	KRT5 and KRT14 Mutations in Epidermolysis Bullosa Simplex with Phenotypic Heterogeneity, and Evidence of Semidominant Inheritance in a Multiplex Family. Journal of Investigative Dermatology, 2016, 136, 1897-1901.	0.7	19
97	Genotypic Heterogeneity and the Mode of Inheritance in Epidermolysis Bullosa. JAMA Dermatology, 2016, 152, 517.	4.1	11
98	Molecular Genetics of the PI3K-AKT-mTOR Pathway in Genodermatoses: Diagnostic Implications and Treatment Opportunities. Journal of Investigative Dermatology, 2016, 136, 15-23.	0.7	35
99	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
100	Lipoid proteinosis: phenotypic heterogeneity in Iranian families with c.507delT mutation in <scp>ECM</scp>1. Experimental Dermatology, 2015, 24, 220-222.	2.9	20
101	The Kindler Syndrome: A Spectrum of FERMT1 Mutations in Iranian Families. Journal of Investigative Dermatology, 2015, 135, 1447-1450.	0.7	26
102	Infantile systemic hyalinosis in an Iranian family with a mutation in theCMG2/ANTXR2gene. Clinical and Experimental Dermatology, 2015, 40, 636-639.	1.3	7
103	Fibroadipose Hyperplasia versus Proteus Syndrome: Segmental Overgrowth with a Mosaic Mutation in the PIK3CA Gene. Journal of Investigative Dermatology, 2015, 135, 1450-1453.	0.7	20
104	BMI1 and TWIST1 Downregulated mRNA Expression in Basal Cell Carcinoma. Asian Pacific Journal of Cancer Prevention, 2014, 15, 3797-3800.	1.2	6
105	Modeling breast acini in tissue culture for detection of malignant phenotype reversion to non-malignant phenotype. Iranian Biomedical Journal, 2009, 13, 191-8.	0.7	3