Lu Liu

List of Publications by Year
in descending order

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25 papers	518 citations	840776 11 h-index	677142 22 g-index
25 all docs	25 docs citations	25 times ranked	944 citing authors

#	Article	IF	Citations
1	Mutations in the ribosome biogenesis factor gene LTV1 are linked to LIPHAK syndrome, a novel poikiloderma-like disorder. Human Molecular Genetics, 2022, 31, 1970-1978.	2.9	4
2	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
3	Griscelli syndrome type 3 in Ethiopian sisters resulting from a homozygous missense mutation in MLPH. International Journal of Dermatology, 2020, 59, e55-e57.	1.0	2
4	Phase I/II open-label trial of intravenous allogeneic mesenchymal stromal cell therapy in adults with recessive dystrophic epidermolysis bullosa. Journal of the American Academy of Dermatology, 2020, 83, 447-454.	1.2	50
5	Homozygous Nonsense Mutation in DSC3 Resulting in Skin Fragility and Hypotrichosis. Journal of Investigative Dermatology, 2020, 140, 1285-1288.	0.7	8
6	Arrhythmogenic right ventricular cardiomyopathy in patients with biallelic JUP-associated skin fragility. Scientific Reports, 2020, 10, 21622.	3.3	7
7	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
8	Ectodermal dysplasiaâ€skin fragility syndrome: Two new cases and review of this desmosomal genodermatosis. Experimental Dermatology, 2020, 29, 520-530.	2.9	4
9	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
10	Semidominant GPNMB Mutations in Amyloidosis Cutis Dyschromica. Journal of Investigative Dermatology, 2019, 139, 2550-2554.e9.	0.7	12
11	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
12	Safety and early efficacy outcomes for lentiviral fibroblast gene therapy in recessive dystrophic epidermolysis bullosa. JCI Insight, 2019, 4, .	5.0	56
13	Homozygous acceptor splice site mutation in DSG1 disrupts plakoglobin localization and results in keratoderma and skin fragility. Journal of Dermatological Science, 2018, 89, 198-201.	1.9	14
14	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
15	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
16	Clinical subtypes and molecular basis of epidermolysis bullosa in Kuwait. International Journal of Dermatology, 2018, 57, 1058-1067.	1.0	10
17	Alopecia, palmoplantar keratoderma, skin fragility and follicular hyperkeratoses due to compound heterozygous mutations in desmoplakin. Australasian Journal of Dermatology, 2017, 58, e17-e19.	0.7	3
18	Mutations in KLHL24 Add to the Molecular Heterogeneity of Epidermolysis BullosaÂSimplex. Journal of Investigative Dermatology, 2017, 137, 1378-1380.	0.7	37

#	Article	IF	Citations
19	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. American Journal of Human Genetics, 2017, 100, 364-370.	6.2	32
20	PLACK syndrome resulting from a new homozygous insertion mutation in CAST. Journal of Dermatological Science, 2017, 88, 256-258.	1.9	10
21	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in aÂSpectrum of Keratinization Disorders Associated with Thrombocytopenia. Journal of Investigative Dermatology, 2017, 137, 2344-2353.	0.7	53
22	Improved molecular diagnosis of the common recurrent intragenic deletion mutation in <scp>IKBKG</scp> in a <scp>F</scp>ilipino family with incontinentia pigmenti. Australasian Journal of Dermatology, 2016, 57, 150-153.	0.7	4
23	Ectodermal dysplasiaâ€“skin fragility syndrome resulting from a new atypical homozygous cryptic acceptor splice site mutation in PKP1. Journal of Dermatological Science, 2016, 84, 210-212.	1.9	3
24	Large Intragenic KRT1 Deletion Underlying Atypical Autosomal Dominant KeratinopathicÂIchthyosis. Journal of Investigative Dermatology, 2016, 136, 2095-2098.	0.7	8
25	Mutations in GRHL2 Result in an Autosomal-Recessive Ectodermal Dysplasia Syndrome. American Journal of Human Genetics, 2014, 95, 308-314.	6.2	48