Henny H Lemmink

List of Publications by Citations

19 papers	160 citations	9 h-index	12 g-index
20 ext. papers	253 ext. citations	5 avg, IF	2.34 L-index

#	Paper	IF	Citations
19	Mutation in exon 1a of PLEC, leading to disruption of plectin isoform 1a, causes autosomal-recessive skin-only epidermolysis bullosa simplex. *Human Molecular Genetics*, **2015**, 24, 3155-62	5.6	34
18	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
17	Germline AGO2 mutations impair RNA interference and human neurological development. *Nature Communications*, **2020**, 11, 5797	17.4	14
16	Intragenic and structural variation in the locus and clinical variability in spinal muscular atrophy. *Brain Communications*, **2020**, 2, fcaa075	4.5	14
15	Association of Epidermolysis Bullosa Simplex With Mottled Pigmentation and EXPH5 Mutations. *JAMA Dermatology*, **2016**, 152, 1137-1141	5.1	14
14	Carriers with functional null mutations in LAMA3 have localized enamel abnormalities due to haploinsufficiency. *European Journal of Human Genetics*, **2016**, 25, 94-99	5.3	11
13	Generalized Ichthyotic Peeling Skin Syndrome due to FLG2 Mutations. *Journal of Investigative Dermatology*, **2018**, 138, 1881-1884	4.3	10
12	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
11	Analysis of , , and as potential disease severity modifiers in spinal muscular atrophy. *Neurology: Genetics*, **2020**, 6, e386	3.8	9
10	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
9	Validation of a Fast, Robust, Inexpensive, Two-Tiered Neonatal Screening Test algorithm on Dried Blood Spots for Spinal Muscular Atrophy. *International Journal of Neonatal Screening*, **2019**, 5, 21	2.6	5
8	Hyperkeratotic hand eczema: Eczema or not?. *Contact Dermatitis*, **2020**, 83, 196-205	2.7	5
7	Genotype-phenotype correlation at codon 1740 of SETD2. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2037-2048	2.5	4
6	A PLEC Isoform Identified in Skin, Muscle, and Heart. *Journal of Investigative Dermatology*, **2017**, 137, 518-522	4.3	3
5	Neurodegenerative VPS41 variants inhibit HOPS function and mTORC1-dependent TFEB/TFE3 regulation. *EMBO Molecular Medicine*, **2021**, 13, e13258	12	3
4	Late onset cardiomyopathy as presenting sign of ATTR A45G amyloidosis caused by a novel TTR mutation (p.A65G). *Cardiovascular Pathology*, **2017**, 29, 19-22	3.8	0
3	Multisite Evaluation and Validation of a Sensitive Diagnostic and Screening System for Spinal Muscular Atrophy that Reports SMN1 and SMN2 Copy Number, along with Disease Modifier and Gene Duplication Variants. *Journal of Molecular Diagnostics*, **2021**, 23, 753-764	5.1	0

2	Feasibility of Follow-Up Studies and Reclassification in Spinocerebellar Ataxia Gene Variants of Unknown Significance.. *Frontiers in Genetics*, **2022**, 13, 782685	4.5	0
1	Deciduous Teeth as an Alternative DNA Source for Postmortem Genetic Testing. *Circulation Genomic and Precision Medicine*, **2020**, 13, e002674	5.2	