Anniek Corveleyn

List of Publications by Year
in descending order

29	1,112	623734	552781
papers	citations	14 h-index	26 g-index

30	30	30	2559
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	2.8	389
2	Disease-associated mutations identify a novel region in human STING necessary for the control of type I interferon signaling. Journal of Allergy and Clinical Immunology, 2017, 140, 543-552.e5.	2.9	159
3	Inherited p40phox deficiency differs from classic chronic granulomatous disease. Journal of Clinical Investigation, 2018, 128, 3957-3975.	8.2	99
4	Mutations in MAGT1 lead to a glycosylation disorder with a variable phenotype. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 9865-9870.	7.1	66
5	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. European Journal of Human Genetics, 2021, 29, 1186-1197.	2.8	61
6	Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154.	4.5	49
7	Adult-Onset ANCA-Associated Vasculitis in SAVI: Extension of the Phenotypic Spectrum, Case Report and Review of the Literature. Frontiers in Immunology, 2020, 11, 575219.	4.8	32
8	Homozygous loss-of-function mutation in ALMS1 causes the lethal disorder mitogenic cardiomyopathy in two siblings. European Journal of Medical Genetics, 2014, 57, 532-535.	1.3	31
9	Human DOCK2 Deficiency: Report of a Novel Mutation and Evidence for Neutrophil Dysfunction. Journal of Clinical Immunology, 2019, 39, 298-308.	3.8	31
10	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	2.8	30
11	PID in Disguise: Molecular Diagnosis of IRAK-4 Deficiency in an Adult Previously Misdiagnosed With Autosomal Dominant Hyper IgE Syndrome. Journal of Clinical Immunology, 2015, 35, 739-744.	3.8	22
12	Successful hematopoietic stem cell transplantation for myelofibrosis in an adult with warts-hypogammaglobulinemia-immunodeficiency-myelokathexis syndrome. Journal of Allergy and Clinical Immunology, 2016, 138, 1485-1489.e2.	2.9	21
13	Individualized corrected QT interval is superior to QT interval corrected using the Bazett formula in predicting mutation carriage in families with long QT syndrome. Heart Rhythm, 2017, 14, 376-382.	0.7	18
14	Compound heterozygous loss-of-function mutations in KIF20A are associated with a novel lethal congenital cardiomyopathy in two siblings. PLoS Genetics, 2018, 14, e1007138.	3.5	18
15	Massive parallel sequencing identifies RAPSN and PDHA1 mutations causing fetal akinesia deformation sequence. European Journal of Paediatric Neurology, 2017, 21, 745-753.	1.6	15
16	Left ventricular non-compaction with Ebstein anomaly attributed to a TPM1 mutation. European Journal of Medical Genetics, 2018, 61, 8-10.	1.3	12
17	Next-generation sequencing in prenatal setting: Some examples of unexpected variant association. European Journal of Medical Genetics, 2020, 63, 103875.	1.3	10
18	Repeat genetic testing with targeted capture sequencing in primary arrhythmia syndrome and cardiomyopathy. European Journal of Human Genetics, 2017, 25, 1313-1323.	2.8	9

#	Article	IF	Citations
19	Clinical and ECG variables to predict the outcome of genetic testing in hypertrophic cardiomyopathy. European Journal of Medical Genetics, 2020, 63, 103754.	1.3	9
20	BCAP31-related syndrome: The first de novo report. European Journal of Medical Genetics, 2020, 63, 103732.	1.3	7
21	A Novel Kindred with MyD88 Deficiency. Journal of Clinical Immunology, 2022, 42, 885-888.	3.8	7
22	Genotypeâ€“phenotype relationship and risk stratification in lossâ€ofâ€function SCN 5A mutation carriers. Annals of Noninvasive Electrocardiology, 2018, 23, e12548.	1.1	6
23	Pathogenic TLR3 Variant in a Patient with Recurrent Herpes Simplex Virus 1â€“Triggered Erythema Multiforme. Journal of Clinical Immunology, 2021, 41, 280-282.	3.8	4
24	Clinical characterization of the first Belgian SCN5A founder mutation cohort. Europace, 2021, 23, 918-927.	1.7	3
25	Targeted capture sequencing in a large LQTS family reveals a new pathogenic mutation c.2038delG in KCNH2 initially missed due to allelic dropout. Acta Cardiologica, 2015, 70, 747-749.	0.9	2
26	Pathogenic P554S Variant in TLR3 in a Patient with Severe Influenza Pneumonia. Journal of Clinical Immunology, 2022, 42, 430-432.	3.8	2
27	P1799GENOTYPE-PHENOTYPE CORRELATION IN A PEDIATRIC ADPKD COHORT. Nephrology Dialysis Transplantation, 2020, 35, .	0.7	0
28	Frequency of Participation in External Quality Assessment Programs Focused on Rare Diseases: Belgian Guidelines for Human Genetics Centers. JMIR Medical Informatics, 2021, 9, e27980.	2.6	0
29	A double-edged sword. Breathe, 2020, 16, 200017.	1.3	0