Marcel Mam Mannens

List of Publications by Year
in descending order

67	6,482	76196	106150
papers	citations	40 h-index	65 g-index

70	70	70	7424
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutation in the KCNQ1 Gene Leading to the Short QT-Interval Syndrome. Circulation, 2004, 109, 2394-2397.	1.6	603
2	Cardiac conduction defects associate with mutations in SCN5A. Nature Genetics, 1999, 23, 20-21.	9.4	549
3	The Human Chitotriosidase Gene. Journal of Biological Chemistry, 1998, 273, 25680-25685.	1.6	360
4	Absence of Calsequestrin 2 Causes Severe Forms of Catecholaminergic Polymorphic Ventricular Tachycardia. Circulation Research, 2002, 91, e21-6.	2.0	358
5	Plakophilin-2 Mutations Are the Major Determinant of Familial Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy. Circulation, 2006, 113, 1650-1658.	1.6	326
6	The RYR2-Encoded Ryanodine Receptor/Calcium Release Channel in Patients Diagnosed Previously With Either Catecholaminergic Polymorphic Ventricular Tachycardia or Genotype Negative, Exercise-Induced Long QT Syndrome. Journal of the American College of Cardiology, 2009, 54, 2065-2074.	1.2	303
7	Compound Heterozygosity for Mutations (W156X and R225W) inSCN5AAssociated With Severe Cardiac Conduction Disturbances and Degenerative Changes in the Conduction System. Circulation Research, 2003, 92, 159-168.	2.0	222
8	Expanding Spectrum of Human RYR2 -Related Disease. Circulation, 2007, 116, 1569-1576.	1.6	211
9	Aniridia-associated cytogenetic rearrangements suggest that a position effect may cause the mutant phenotype. Human Molecular Genetics, 1995, 4, 415-422.	1.4	195
10	Hypomethylation of the H19 Gene Causes Not Only Silver-Russell Syndrome (SRS) but Also Isolated Asymmetry or an SRS-Like Phenotype. American Journal of Human Genetics, 2006, 78, 604-614.	2.6	186
11	Human SCN5A gene mutations alter cardiac sodium channel kinetics and are associated with the Brugada syndrome. Cardiovascular Research, 1999, 44, 507-517.	1.8	181
12	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	2.6	171
13	Evidence that WT1 mutations in Denys â€" Drash syndrome patients may act in a dominant-negative fashion. Human Molecular Genetics, 1993, 2, 259-264.	1.4	158
14	Haplotype-Sharing Analysis Implicates Chromosome 7q36 Harboring DPP6 in Familial IdiopathicÂVentricular Fibrillation. American Journal of Human Genetics, 2009, 84, 468-476.	2.6	158
15	Allelic loss of chromosome 1p36 in neuroblastoma is of preferential maternal origin and correlates with Nâ€“myc amplification. Nature Genetics, 1993, 4, 187-190.	9.4	147
16	Yield of Molecular and Clinical Testing for Arrhythmia Syndromes. Circulation, 2013, 128, 1513-1521.	1.6	132
17	The 2373insG mutation in the MYBPC3 gene is a founder mutation, which accounts for nearly one-fourth of the HCM cases in the Netherlands. European Heart Journal, 2003, 24, 1848-1853.	1.0	127
18	Truncating titin mutations are associated with a mild and treatable form of dilated cardiomyopathy. European Journal of Heart Failure, 2017, 19, 512-521.	2.9	127

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19	Genetics of Beckwith-Wiedemann syndrome-associated tumors: Common genetic pathways. Genes Chromosomes and Cancer, 2000, 28, 1-13.	1.5	101
20	Lessons from BWS twins: complex maternal and paternal hypomethylation and a common source of haematopoietic stem cells. European Journal of Human Genetics, 2009, 17, 1625-1634.	1.4	98
21	Diagnostic criteria for congenital long QT syndrome in the era of molecular genetics: do we need a scoring system?. European Heart Journal, 2006, 28, 575-580.	1.0	96
22	Epigenotyping as a tool for the prediction of tumor risk and tumor type in patients with Beckwith-Wiedemann syndrome (BWS). Journal of Pediatrics, 2004, 145, 796-799.	0.9	93
23	A Dominant Negative Isoform of the Long QT Syndrome 1 Gene Product. Journal of Biological Chemistry, 1998, 273, 6837-6843.	1.6	82
24	The Human Achaete-Scute Homologue 2 (ASCL2, HASH2) Maps to Chromosome 11p15.5, Close to IGF2 and is Expressed in Extravillus Trophoblasts. Human Molecular Genetics, 1997, 6, 859-867.	1.4	79
25	Desmoglein-2 and Desmocollin-2 Mutations in Dutch Arrhythmogenic Right Ventricular Dysplasia/Cardiomypathy Patients. Circulation: Cardiovascular Genetics, 2009, 2, 418-427.	5.1	77
26	A Novel Early Onset Lethal Form of Catecholaminergic Polymorphic Ventricular Tachycardia Maps to Chromosome 7p14-p22. Journal of Cardiovascular Electrophysiology, 2007, 18, 1060-1066.	0.8	74
27	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387.	1.4	68
28	Homozygous Premature Truncation of the HERG Protein. Circulation, 1999, 100, 1264-1267.	1.6	67
29	Long QT syndrome caused by a large duplication in the KCNH2 (HERG) gene undetectable by current polymerase chain reaction-based exon-scanning methodologies. Heart Rhythm, 2006, 3, 52-55.	0.3	65
30	A high-resolution integrated physical, cytogenetic, and genetic map of human chromosome 11: distal p13 to proximal p15.1. Genomics, 1995, 25, 447-461.	1.3	58
31	Recurrent intrauterine fetal loss due to near absence of HERG: Clinical and functional characterization of a homozygous nonsense HERG Q1070X mutation. Heart Rhythm, 2008, 5, 553-561.	0.3	58
32	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. European Journal of Human Genetics, 2015, 23, 1142-1150.	1.4	56
33	An epigenome-wide association study in whole blood of measures of adiposity among Ghanaians: the RODAM study. Clinical Epigenetics, 2017, 9, 103.	1.8	55
34	Characterization of regions of chromosomes 12 and 16 involved in nephroblastoma tumorigenesis. Genes Chromosomes and Cancer, 1995, 14, 285-294.	1.5	50
35	Clinical utility gene card for: Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	1.4	50
36	Exclusion of multiple candidate genes and large genomic rearrangements in SCN5A in a Dutch Brugada syndrome cohort. Heart Rhythm, 2007, 4, 752-755.	0.3	48

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37	Determination of KCNQ1OT1 and H19 methylation levels in BWS and SRS patients using methylation-sensitive high-resolution melting analysis. European Journal of Human Genetics, 2009, 17, 467-473.	1.4	47
38	An intronic mutation leading to incomplete skipping of exon-2 in KCNQ1 rescues hearing in Jervell and Lange-Nielsen syndrome. Progress in Biophysics and Molecular Biology, 2008, 98, 319-327.	1.4	44
39	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	1.4	44
40	Molecular genetic testing for familial hypercholesterolemia: spectrum of LDL receptor gene mutations in the Netherlands. Clinical Genetics, 2000, 57, 116-124.	1.0	43
41	An inactivating mutation in the histone deacetylase SIRT6 causes human perinatal lethality. Genes and Development, 2018, 32, 373-388.	2.7	41
42	Arrhythmogenic right ventricular cardiomyopathy due to a novel plakophilin 2 mutation: Wide spectrum of disease in mutation carriers within a family. Heart Rhythm, 2006, 3, 939-944.	0.3	40
43	Molecular characterisation of 10 Dutch properdin type I deficient families: mutation analysis and X-inactivation studies. European Journal of Human Genetics, 2000, 8, 513-518.	1.4	39
44	A case of methemoglobinemia type II due to NADH-cytochrome b5 reductase deficiency: Determination of the molecular basis. Human Mutation, 2000, 16, 18-22.	1.1	36
45	The idiopathic preterm delivery methylation profile in umbilical cord blood DNA. BMC Genomics, 2015, 16, 736.	1.2	33
46	Methylation analysis in tongue tissue of BWS patients identifies the (EPI)genetic cause in 3 patients with normal methylation levels in blood. European Journal of Medical Genetics, 2014, 57, 293-297.	0.7	27
47	NTCP deficiency and persistently raised bile salts: an adult case. Journal of Inherited Metabolic Disease, 2017, 40, 313-315.	1.7	27
48	Novel tools for extraction and validation of disease-related mutations applied to fabry disease. Human Mutation, 2010, 31, 1026-1032.	1.1	22
49	Postpacing abnormal repolarization in catecholaminergic polymorphic ventricular tachycardia associated with a mutation in the cardiac ryanodine receptor gene. Heart Rhythm, 2011, 8, 1546-1552.	0.3	22
50	Perceived discrimination and stressful life events are associated with cardiovascular risk score in migrant and non-migrant populations: The RODAM study. International Journal of Cardiology, 2019, 286, 169-174.	0.8	21
51	Prevalence of type 2 diabetes and its association with measures of body composition among African residents in the Netherlands â€“ The HELIUS study. Diabetes Research and Clinical Practice, 2015, 110, 137-146.	1.1	20
52	Prevalence and determinants of type 2 diabetes among lean African migrants and non-migrants: the RODAM study. Journal of Global Health, 2019, 9, 020426.	1.2	20
53	Efficient Molecular Diagnostic Strategy for ABCC6 in Pseudoxanthoma Elasticum. Genetic Testing and Molecular Biomarkers, 2004, 8, 292-300.	1.7	19
54	Review Article: Genomic imprinting: concept and clinical consequences. Annals of Medicine, 1999, 31, 4-11.	1.5	18

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55	The generation of ordered sets of cosmid DNA clones from human chromosome region 11p. Genomics, 1992, 13, 89-94.	1.3	16
56	KVLQT1, the rhythm of imprinting. Nature Genetics, 1997, 15, 113-115.	9.4	16
57	Cytogenetic and molecular analysis of cellular atypical mesoblastic nephroma. , 1998, 21, 265-269.		16
58	Identification of copy number variants associated with BPES-like phenotypes. Human Genetics, 2008, 124, 489-498.	1.8	15
59	Detection of a cryptic paracentric inversion within band 11p13 in familial aniridia by fluorescence in situ hybridization. Human Genetics, 1993, 91, 205-9.	1.8	13
60	Pericentric intrachromosomal insertion responsible for recurrence of del(11)(p13p14) in a family. Genes Chromosomes and Cancer, 1993, 7, 57-62.	1.5	12
61	Differential DNA Methylation Is Associated With Hippocampal Abnormalities in Pediatric Posttraumatic Stress Disorder. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2021, 6, 1063-1070.	1.1	8
62	DNA methylation as the link between migration and theÂmajor noncommunicable diseases: the RODAM study. Epigenomics, 2021, 13, 653-666.	1.0	5
63	Genome-wide DNA methylation analysis on C-reactive protein among Ghanaians suggests molecular links to the emerging risk of cardiovascular diseases. Npj Genomic Medicine, 2021, 6, 46.	1.7	4
64	Screening for inborn errors of metabolism in psychotic patients using Next Generation Sequencing. Journal of Psychiatric Research, 2021, 138, 125-129.	1.5	1
65	Prenatal NeuN+ neurons of Down syndrome display aberrant integrative DNA methylation and gene expression profiles. Epigenomics, 2022, 14, 375-390.	1.0	1
66	Tumor suppressor genes in neuroblastoma. Cancer Genetics and Cytogenetics, 1992, 63, 169.	1.0	0
67	The human genome project: Deciphering the blueprint of heredity. Trends in Genetics, 1995, 11, 418-419.	2.9	0