

# Marcel Mam Mannens

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/1022792/publications.pdf>

Version: 2024-02-01

67  
papers

6,482  
citations

76294

40  
h-index

106281

65  
g-index

70  
all docs

70  
docs citations

70  
times ranked

7424  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutation in the KCNQ1 Gene Leading to the Short QT-Interval Syndrome. <i>Circulation</i> , 2004, 109, 2394-2397.	1.6	603
2	Cardiac conduction defects associate with mutations in SCN5A. <i>Nature Genetics</i> , 1999, 23, 20-21.	9.4	549
3	The Human Chitotriosidase Gene. <i>Journal of Biological Chemistry</i> , 1998, 273, 25680-25685.	1.6	360
4	Absence of Calsequestrin 2 Causes Severe Forms of Catecholaminergic Polymorphic Ventricular Tachycardia. <i>Circulation Research</i> , 2002, 91, e21-6.	2.0	358
5	Plakophilin-2 Mutations Are the Major Determinant of Familial Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy. <i>Circulation</i> , 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. <i>Journal of the American College of Cardiology</i> , 2009, 54, 2065-2074.	1.2	303
7	Compound Heterozygosity for Mutations (W156X and R225W) in SCN5A Associated With Severe Cardiac Conduction Disturbances and Degenerative Changes in the Conduction System. <i>Circulation Research</i> , 2003, 92, 159-168.	2.0	222
8	Expanding Spectrum of Human <i>RYR2</i> -Related Disease. <i>Circulation</i> , 2007, 116, 1569-1576.	1.6	211
9	Aniridia-associated cytogenetic rearrangements suggest that a position effect may cause the mutant phenotype. <i>Human Molecular Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2006, 78, 604-614.	2.6	186
11	Human SCN5A gene mutations alter cardiac sodium channel kinetics and are associated with the Brugada syndrome. <i>Cardiovascular Research</i> , 1999, 44, 507-517.	1.8	181
12	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 356-370.	2.6	171
13	Evidence that WT1 mutations in Denys-Drash syndrome patients may act in a dominant-negative fashion. <i>Human Molecular Genetics</i> , 1993, 2, 259-264.	1.4	158
14	Haplotype-Sharing Analysis Implicates Chromosome 7q36 Harboring DPP6 in Familial Idiopathic Ventricular Fibrillation. <i>American Journal of Human Genetics</i> , 2009, 84, 468-476.	2.6	158
15	Allelic loss of chromosome 1p36 in neuroblastoma is of preferential maternal origin and correlates with <i>myc</i> amplification. <i>Nature Genetics</i> , 1993, 4, 187-190.	9.4	147
16	Yield of Molecular and Clinical Testing for Arrhythmia Syndromes. <i>Circulation</i> , 2013, 128, 1513-1521.	1.6	132
17	The 2373insC mutation in the MYBPC3 gene is a founder mutation, which accounts for nearly one-fourth of the HCM cases in the Netherlands. <i>European Heart Journal</i> , 2003, 24, 1848-1853.	1.0	127
18	Truncating titin mutations are associated with a mild and treatable form of dilated cardiomyopathy. <i>European Journal of Heart Failure</i> , 2017, 19, 512-521.	2.9	127

#	ARTICLE	IF	CITATIONS
19	Genetics of Beckwith-Wiedemann syndrome-associated tumors: Common genetic pathways. <i>Genes Chromosomes and Cancer</i> , 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. <i>European Journal of Human Genetics</i> , 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?. <i>European Heart Journal</i> , 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). <i>Journal of Pediatrics</i> , 2004, 145, 796-799.	0.9	93
23	A Dominant Negative Isoform of the Long QT Syndrome 1 Gene Product. <i>Journal of Biological Chemistry</i> , 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 Extravillous Trophoblasts. <i>Human Molecular Genetics</i> , 1997, 6, 859-867.	1.4	79
25	Desmoglein-2 and Desmocollin-2 Mutations in Dutch Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Patients. <i>Circulation: Cardiovascular Genetics</i> , 2009, 2, 418-427.	5.1	77
26	A Novel Early Onset Lethal Form of Catecholaminergic Polymorphic Ventricular Tachycardia Maps to Chromosome 7p14-p22. <i>Journal of Cardiovascular Electrophysiology</i> , 2007, 18, 1060-1066.	0.8	74
27	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silver-Russell and Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1377-1387.	1.4	68
28	Homozygous Premature Truncation of the HERG Protein. <i>Circulation</i> , 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. <i>Heart Rhythm</i> , 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. <i>Genomics</i> , 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. <i>Heart Rhythm</i> , 2008, 5, 553-561.	0.3	58
32	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. <i>European Journal of Human Genetics</i> , 2015, 23, 1142-1150.	1.4	56
33	An epigenome-wide association study in whole blood of measures of adiposity among Chanaians: the RODAM study. <i>Clinical Epigenetics</i> , 2017, 9, 103.	1.8	55
34	Characterization of regions of chromosomes 12 and 16 involved in nephroblastoma tumorigenesis. <i>Genes Chromosomes and Cancer</i> , 1995, 14, 285-294.	1.5	50
35	Clinical utility gene card for: Beckwith-Wiedemann Syndrome. <i>European Journal of Human Genetics</i> , 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. <i>Heart Rhythm</i> , 2007, 4, 752-755.	0.3	48

#	ARTICLE	IF	CITATIONS
37	Determination of KCNQ1OT1 and H19 methylation levels in BWS and SRS patients using methylation-sensitive high-resolution melting analysis. <i>European Journal of Human Genetics</i> , 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. <i>Progress in Biophysics and Molecular Biology</i> , 2008, 98, 319-327.	1.4	44
39	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. <i>European Journal of Human Genetics</i> , 2016, 24, 784-793.	1.4	44
40	Molecular genetic testing for familial hypercholesterolemia: spectrum of LDL receptor gene mutations in the Netherlands. <i>Clinical Genetics</i> , 2000, 57, 116-124.	1.0	43
41	An inactivating mutation in the histone deacetylase SIRT6 causes human perinatal lethality. <i>Genes and Development</i> , 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. <i>Heart Rhythm</i> , 2006, 3, 939-944.	0.3	40
43	Molecular characterisation of 10 Dutch properdin type I deficient families: mutation analysis and X-inactivation studies. <i>European Journal of Human Genetics</i> , 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. <i>Human Mutation</i> , 2000, 16, 18-22.	1.1	36
45	The idiopathic preterm delivery methylation profile in umbilical cord blood DNA. <i>BMC Genomics</i> , 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. <i>European Journal of Medical Genetics</i> , 2014, 57, 293-297.	0.7	27
47	NTCP deficiency and persistently raised bile salts: an adult case. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 313-315.	1.7	27
48	Novel tools for extraction and validation of disease-related mutations applied to fabry disease. <i>Human Mutation</i> , 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. <i>Heart Rhythm</i> , 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. <i>International Journal of Cardiology</i> , 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 – The HELIUS study. <i>Diabetes Research and Clinical Practice</i> , 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. <i>Journal of Global Health</i> , 2019, 9, 020426.	1.2	20
53	Efficient Molecular Diagnostic Strategy for ABCC6 in Pseudoxanthoma Elasticum. <i>Genetic Testing and Molecular Biomarkers</i> , 2004, 8, 292-300.	1.7	19
54	Review Article: Genomic imprinting: concept and clinical consequences. <i>Annals of Medicine</i> , 1999, 31, 4-11.	1.5	18

#	ARTICLE	IF	CITATIONS
55	The generation of ordered sets of cosmid DNA clones from human chromosome region 11p. <i>Genomics</i> , 1992, 13, 89-94.	1.3	16
56	KVLQT1, the rhythm of imprinting. <i>Nature Genetics</i> , 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. <i>Human Genetics</i> , 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. <i>Human Genetics</i> , 1993, 91, 205-9.	1.8	13
60	Pericentric intrachromosomal insertion responsible for recurrence of del(11)(p13p14) in a family. <i>Genes Chromosomes and Cancer</i> , 1993, 7, 57-62.	1.5	12
61	Differential DNA Methylation Is Associated With Hippocampal Abnormalities in Pediatric Posttraumatic Stress Disorder. <i>Biological Psychiatry: Cognitive Neuroscience and Neuroimaging</i> , 2021, 6, 1063-1070.	1.1	8
62	DNA methylation as the link between migration and the major noncommunicable diseases: the RODAM study. <i>Epigenomics</i> , 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. <i>Npj Genomic Medicine</i> , 2021, 6, 46.	1.7	4
64	Screening for inborn errors of metabolism in psychotic patients using Next Generation Sequencing. <i>Journal of Psychiatric Research</i> , 2021, 138, 125-129.	1.5	1
65	Prenatal NeuN+ neurons of Down syndrome display aberrant integrative DNA methylation and gene expression profiles. <i>Epigenomics</i> , 2022, 14, 375-390.	1.0	1
66	Tumor suppressor genes in neuroblastoma. <i>Cancer Genetics and Cytogenetics</i> , 1992, 63, 169.	1.0	0
67	The human genome project: Deciphering the blueprint of heredity. <i>Trends in Genetics</i> , 1995, 11, 418-419.	2.9	0