Mieke M Van Haelst

List of Publications by Year
in descending order

60 papers	1,980 citations	304743 22 h-index	276875 41 g-index
61 all docs	61 docs citations	61 times ranked	4638 citing authors

#	Article	IF	Citations
1	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
2	Dominant missense mutations in ABCC9 cause CantÃº syndrome. Nature Genetics, 2012, 44, 793-796.	21.4	184
3	Effectiveness of whole-exome sequencing and costs of the traditional diagnostic trajectory in children with intellectual disability. Genetics in Medicine, 2016, 18, 949-956.	2.4	148
4	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	2.5	116
5	Drug Repurposing for Rare Diseases. Trends in Pharmacological Sciences, 2021, 42, 255-267.	8.7	105
6	Lysosomal Signaling Licenses Embryonic Stem Cell Differentiation via Inactivation of Tfe3. Cell Stem Cell, 2019, 24, 257-270.e8.	11.1	97
7	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. Genetics in Medicine, 2021, 23, 1065-1074.	2.4	88
8	Truncating Homozygous Mutation of Carboxypeptidase E (CPE) in a Morbidly Obese Female with Type 2 Diabetes Mellitus, Intellectual Disability and Hypogonadotrophic Hypogonadism. PLoS ONE, 2015, 10, e0131417.	2.5	72
9	Effective CRISPR/Cas9-based nucleotide editing in zebrafish to model human genetic cardiovascular disorders. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	69
10	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. European Journal of Human Genetics, 2015, 23, 1176-1185.	2.8	67
11	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. American Journal of Human Genetics, 2016, 98, 963-970.	6.2	67
12	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	3.8	66
13	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	6.2	61
14	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	6.2	53
15	Leptin receptor deficiency: a systematic literature review and prevalence estimation based on population genetics. European Journal of Endocrinology, 2020, 182, 47-56.	3.7	51
16	CantÃº syndrome: Findings from 74 patients in the International CantÃº Syndrome Registry. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 658-681.	1.6	50
17	Further confirmation of the MED13L haploinsufficiency syndrome. European Journal of Human Genetics, 2015, 23, 135-138.	2.8	40
18	Non-invasive sources of cells with primary cilia from pediatric and adult patients. Cilia, 2015, 4, 8.	1.8	34

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19	Fetal methotrexate syndrome: A systematic review of case reports. Reproductive Toxicology, 2019, 87, 125-139.	2.9	33
20	Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations. European Journal of Human Genetics, 2014, 22, 652-659.	2.8	32
21	Ankyrin repeat and zinc-finger domain-containing 1 mutations are associated with infantile-onset inflammatory bowel disease. Journal of Biological Chemistry, 2017, 292, 7904-7920.	3.4	29
22	Identifying underlying medical causes of pediatric obesity: Results of a systematic diagnostic approach in a pediatric obesity center. PLoS ONE, 2020, 15, e0232990.	2.5	28
23	Whole-exome sequencing in pediatrics: parentsâ€™ considerations toward return of unsolicited findings for their child. European Journal of Human Genetics, 2016, 24, 1681-1687.	2.8	22
24	Bariatric Surgery for Monogenic Non-syndromic and Syndromic Obesity Disorders. Current Diabetes Reports, 2020, 20, 44.	4.2	20
25	Establishing the phenotypic spectrum of ZTTK syndrome by analysis of 52 individuals with variants in SON. European Journal of Human Genetics, 2022, 30, 271-281.	2.8	19
26	Second case of Bardetâ€“Biedl syndrome caused by biallelic variants in IFT74. European Journal of Human Genetics, 2020, 28, 943-946.	2.8	18
27	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
28	Long-Term Weight Outcome After Bariatric Surgery in Patients with Melanocortin-4 Receptor Gene Variants: a Caseâ€“Control Study of 105 Patients. Obesity Surgery, 2022, 32, 837-844.	2.1	15
29	Expanding the phenotype of biallelic <scp>RNPC3</scp> variants associated with growth hormone deficiency. American Journal of Medical Genetics, Part A, 2020, 182, 1952-1956.	1.2	14
30	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
31	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
32	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
33	Functional Analysis of the SIM1 Variant p.G715V in 2 Patients With Obesity. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 355-361.	3.6	11
34	Effects of <scp>glucagonâ€like</scp> peptideâ€1 analogue treatment in genetic obesity: A case series. Clinical Obesity, 2021, 11, e12481.	2.0	11
35	Biallelic BUB1 mutations cause microcephaly, developmental delay, and variable effects on cohesion and chromosome segregation. Science Advances, 2022, 8, eabk0114.	10.3	11
36	Who ever heard of 16p11.2 deletion syndrome? Parentsâ€™ perspectives on a susceptibility copy number variation syndrome. European Journal of Human Genetics, 2020, 28, 1196-1204.	2.8	10

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37	4H leukodystrophy caused by a homozygous POLR3B mutation: Further delineation of the phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 1776-1779.	1.2	10
38	CantÃº syndrome, the changing phenotype: a report of the two oldest Dutch patients. Clinical Dysmorphology, 2018, 27, 78-83.	0.3	9
39	PDE3A gene screening improves diagnostics for patients with Bilginturan syndrome (hypertension and) Tj ETQq1 1 0.784314 rgBT /Ov	2.7	9
40	Mosaic CREBBP mutation causes overlapping clinical features of Rubinsteinâ€“Taybi and Filippi syndromes. European Journal of Human Genetics, 2016, 24, 1363-1366.	2.8	8
41	Expanding the spectrum of CEP55â€associated disease to viable phenotypes. American Journal of Medical Genetics, Part A, 2020, 182, 1201-1208.	1.2	8
42	Threeâ€dimensional facial morphology in CantÃº syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1041-1052.	1.2	8
43	Clinical utility gene card for: CantÃº syndrome. European Journal of Human Genetics, 2017, 25, 512-512.	2.8	5
44	Genetic analysis in the bariatric clinic; impact of a <scp>PTEN</scp> gene mutation. Molecular Genetics & Genomic Medicine, 2019, 7, e00632.	1.2	4
45	The Diagnostic Journey of a Patient with Praderâ€“Willi-Like Syndrome and a Unique Homozygous SNURF-SNRPN Variant; Bio-Molecular Analysis and Review of the Literature. Genes, 2021, 12, 875.	2.4	4
46	Genetic diagnosis for rare diseases in the Dutch Caribbean: a qualitative study on the experiences and associated needs of parents. European Journal of Human Genetics, 2022, , .	2.8	4
47	De novo 14q24.2q24.3 microdeletion including IFT43 is associated with intellectual disability, skeletal anomalies, cardiac anomalies, and myopia. American Journal of Medical Genetics, Part A, 2016, 170, 1566-1569.	1.2	3
48	Structural Models for the Dynamic Effects of Loss-of-Function Variants in the Human SIM1 Protein Transcriptional Activation Domain. Biomolecules, 2020, 10, 1314.	4.0	3
49	Behavioral and cognitive functioning in individuals with CantÃº syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2434-2444.	1.2	3
50	Novel SCN9A Mutations in a Compound Heterozygous Girl with Congenital Insensitivity to Pain. Journal of Pediatric Neurology, 2021, 19, 189-192.	0.2	3
51	Limb anomalies, microcephaly, dysmorphic facial features and fibroma of the tongue after failed abortion with methotrexate and misoprostol. Clinical Dysmorphology, 2020, 29, 182-185.	0.3	2
52	Genetic care in geographically isolated small island communities: 8Âyears of experience in the Dutch Caribbean. American Journal of Medical Genetics, Part A, 2022, 188, 1777-1791.	1.2	2
53	De NovoTrisomy 1q10q23.3 Mosaicism Causes Microcephaly, Severe Developmental Delay, and Facial Dysmorphic Features but No Cardiac Anomalies. Case Reports in Genetics, 2016, 2016, 1-5.	0.2	1
54	Clinical and community genetics services in the Dutch Caribbean. Journal of Community Genetics, 2021, 12, 497-501.	1.2	1

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55	Corrigendum to Obesity Syndrome, MOMES Caused by Deletion-Duplication (4q35.1 del and 5p14.3) Tj ETQq1 1 0.784314 rgBT /Ove 1874-1874.	1.2	0
56	Young adult with CantÃº syndrome: dealing with a rare genetic skin disorder. BMJ Case Reports, 2021, 14, e243118.	0.5	0
57	Title is missing!. , 2020, 15, e0232990.		0
58	Title is missing!. , 2020, 15, e0232990.		0
59	Title is missing!. , 2020, 15, e0232990.		0
60	Title is missing!. , 2020, 15, e0232990.		0