

Conny M A Van Ravenswaaij-Arts

List of Publications by Year in descending order

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Version: 2024-02-01

59
papers

2,630
citations

201674

27
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206112

48
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61
all docs

61
docs citations

61
times ranked

6110
citing authors

#	ARTICLE	IF	CITATIONS
1	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016, 17, 243.	8.8	241
2	Mutation update on the CHD7 gene involved in CHARGE syndrome. <i>Human Mutation</i> , 2012, 33, 1149-1160.	2.5	224
3	<i>GRIN2A</i> -related disorders: genotype and functional consequence predict phenotype. <i>Brain</i> , 2019, 142, 80-92.	7.6	143
4	<i>SYNGAP1</i> encephalopathy. <i>Neurology</i> , 2019, 92, e96-e107.	1.1	131
5	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) <i>Tj ETQq1 1 0.784314 rgBT /Over</i>	2.8	109
6	Improving the diagnostic yield of exome-sequencing by predicting gene-phenotype associations using large-scale gene expression analysis. <i>Nature Communications</i> , 2019, 10, 2837.	12.8	107
7	De Novo Loss-of-Function Mutations in <i>USP9X</i> Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. <i>American Journal of Human Genetics</i> , 2016, 98, 373-381.	6.2	95
8	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. <i>Genetics in Medicine</i> , 2017, 19, 45-52.	2.4	94
9	Molecular and clinical characterization of 25 individuals with exonic deletions of <i>NRXN1</i> and comprehensive review of the literature. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2013, 162, 388-403.	1.7	93
10	Towards a European consensus for reporting incidental findings during clinical NGS testing. <i>European Journal of Human Genetics</i> , 2015, 23, 1601-1606.	2.8	85
11	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. <i>American Journal of Human Genetics</i> , 2015, 97, 483-492.	6.2	70
12	The Results of <i>CHD7</i> Analysis in Clinically Well-Characterized Patients with Kallmann Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E858-E862.	3.6	69
13	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor <i>SIN3A</i> causes mild intellectual disability by affecting the development of cortical integrity. <i>Nature Genetics</i> , 2016, 48, 877-887.	21.4	67
14	Functional Insights into Chromatin Remodelling from Studies on CHARGE Syndrome. <i>Trends in Genetics</i> , 2015, 31, 600-611.	6.7	66
15	Whole-exome sequencing is a powerful approach for establishing the etiological diagnosis in patients with intellectual disability and microcephaly. <i>BMC Medical Genomics</i> , 2015, 9, 7.	1.5	65
16	The introduction of arrays in prenatal diagnosis: A special challenge. <i>Human Mutation</i> , 2012, 33, 923-929.	2.5	63
17	<i>MYT1L</i> is a candidate gene for intellectual disability in patients with 2p25.3 (2pter) deletions. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2739-2745.	1.2	53
18	Haploinsufficiency of the <i>STX1B</i> gene is associated with myoclonic astatic epilepsy. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 489-492.	1.6	52

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19	Developmental phenotype in Phelan-McDermid (22q13.3 deletion) syndrome: a systematic and prospective study in 34 children. <i>Journal of Neurodevelopmental Disorders</i> , 2016, 8, 16.	3.1	51
20	New insights and advances in CHARGE syndrome: Diagnosis, etiologies, treatments, and research discoveries. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017, 175, 397-406.	1.6	46
21	Anosmia Predicts Hypogonadotropic Hypogonadism in CHARGE Syndrome. <i>Journal of Pediatrics</i> , 2011, 158, 474-479.	1.8	45
22	CHARGE syndrome: a review of the immunological aspects. <i>European Journal of Human Genetics</i> , 2015, 23, 1451-1459.	2.8	44
23	<i>CREBBP</i> mutations in individuals without Rubinstein-Taybi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2681-2693.	1.2	43
24	De Novo Truncating Mutations in <i>WASF1</i> Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	6.2	36
25	Exon copy number alterations of the <i>CHD7</i> gene are not a major cause of CHARGE and CHARGE-like syndrome. <i>European Journal of Medical Genetics</i> , 2008, 51, 417-425.	1.3	33
26	Haploinsufficiency for <i>ANKRD11</i> -flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. <i>European Journal of Human Genetics</i> , 2017, 25, 694-701.	2.8	33
27	Developing a CHARGE syndrome checklist: Health supervision across the lifespan (from head to toe). <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 684-691.	1.2	32
28	Phelan-McDermid syndrome: a classification system after 30 years of experience. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 27.	2.7	32
29	The phenotypic spectrum of proximal 6q deletions based on a large cohort derived from social media and literature reports. <i>European Journal of Human Genetics</i> , 2018, 26, 1478-1489.	2.8	31
30	Is there an effect of intranasal insulin on development and behaviour in Phelan-McDermid syndrome? A randomized, double-blind, placebo-controlled trial. <i>European Journal of Human Genetics</i> , 2016, 24, 1696-1701.	2.8	30
31	Guidelines in CHARGE syndrome and the missing link: Cranial imaging. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017, 175, 450-464.	1.6	29
32	Immune Dysfunction in Children with CHARGE Syndrome: A Cross-Sectional Study. <i>PLoS ONE</i> , 2015, 10, e0142350.	2.5	27
33	Neurodegenerative <i>VPS41</i> variants inhibit HOPS function and mTORC1-dependent TFEB/TFE3 regulation. <i>EMBO Molecular Medicine</i> , 2021, 13, e13258.	6.9	26
34	A novel homozygous insertion and review of published mutations in the <i>NNT</i> gene causing familial glucocorticoid deficiency (FGD). <i>European Journal of Medical Genetics</i> , 2015, 58, 642-649.	1.3	24
35	Loss-of-function and missense variants in <i>NSD2</i> cause decreased methylation activity and are associated with a distinct developmental phenotype. <i>Genetics in Medicine</i> , 2021, 23, 1474-1483.	2.4	24
36	<i>Sema3a</i> plays a role in the pathogenesis of CHARGE syndrome. <i>Human Molecular Genetics</i> , 2018, 27, 1343-1352.	2.9	20

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37	PRRT2-related phenotypes in patients with a 16p11.2 deletion. <i>European Journal of Medical Genetics</i> , 2019, 62, 265-269.	1.3	20
38	TAB2 deletions and variants cause a highly recognisable syndrome with mitral valve disease, cardiomyopathy, short stature and hypermobility. <i>European Journal of Human Genetics</i> , 2021, 29, 1669-1676.	2.8	19
39	Clinical utility gene card for: CHARGE syndrome - update 2015. <i>European Journal of Human Genetics</i> , 2015, 23, 3-4.	2.8	18
40	Influence of hearing loss and cognitive abilities on language development in CHARGE Syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2022-2030.	1.2	14
41	Congenital arch vessel anomalies in CHARGE syndrome: A frequent feature with risk for co-morbidity. <i>IJC Heart and Vasculature</i> , 2016, 12, 21-25.	1.1	14
42	Copy number variation in a hospital-based cohort of children with epilepsy. <i>Epilepsia Open</i> , 2017, 2, 244-254.	2.4	13
43	Suggestions for a Guideline for Cochlear Implantation in CHARGE Syndrome. <i>Otology and Neurotology</i> , 2016, 37, 1275-1283.	1.3	12
44	Duplication 2p25 in a child with clinical features of CHARGE syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1148-1154.	1.2	11
45	<i>CHD</i>7 mutations are not a major cause of atrioventricular septal and conotruncal heart defects. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3003-3009.	1.2	10
46	Imaging of Clival Hypoplasia in CHARGE Syndrome and Hypothesis for Development: A Case-Control Study. <i>American Journal of Neuroradiology</i> , 2018, 39, 1938-1942.	2.4	9
47	A 649kb microduplication in 1p34.1, including POMGNT1, in a patient with microcephaly, coloboma and laryngomalacia; and a review of the literature. <i>European Journal of Medical Genetics</i> , 2009, 52, 116-119.	1.3	8
48	Exome sequencing identifies the first genetic determinants of sirenomelia in humans. <i>Human Mutation</i> , 2020, 41, 926-933.	2.5	8
49	Under-reported aspects of diagnosis and treatment addressed in the Dutch-Flemish guideline for comprehensive diagnostics in disorders/differences of sex development. <i>Journal of Medical Genetics</i> , 2020, 57, 581-589.	3.2	8
50	Cerebellar Vermis and Midbrain Hypoplasia Upon Conditional Deletion of Chd7 from the Embryonic Mid-Hindbrain Region. <i>Frontiers in Neuroanatomy</i> , 2017, 11, 86.	1.7	7
51	Can Characteristics of Reciprocal Translocations Predict the Chance of Transferable Embryos in PGD Cycles?. <i>Journal of Clinical Medicine</i> , 2014, 3, 348-358.	2.4	6
52	Central Adrenal Insufficiency Is Not a Common Feature in CHARGE Syndrome: A Cross-Sectional Study in 2 Cohorts. <i>Journal of Pediatrics</i> , 2016, 176, 150-155.	1.8	5
53	Changes in empowerment and anxiety of patients and parents during genetic counselling for epilepsy. <i>European Journal of Paediatric Neurology</i> , 2021, 32, 128-135.	1.6	4
54	<sc>EPA7</sc> haploinsufficiency is associated with a neurodevelopmental disorder. <i>Clinical Genetics</i> , 2021, 100, 396-404.	2.0	3

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55	Mechanism of Disease: Recessive ADAMTSL4 Mutations and Craniosynostosis with Ectopia Lentis. Case Reports in Genetics, 2022, 2022, 1-8.	0.2	3
56	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	2.8	1
57	An analysis of body proportions in children with CHARGE syndrome using photogrammetric anthropometry. American Journal of Medical Genetics, Part A, 2019, 179, 1459-1465.	1.2	1
58	CHARGE syndrome and related disorders: a mechanistic link. Human Molecular Genetics, 2021, 30, 2215-2224.	2.9	1
59	Support for the Diagnosis of CHARGE Syndrome. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 634.	2.2	0