Conny M A Van Ravenswaaij-Arts

List of Publications by Year
in descending order

59 papers	2,630 citations	201674 27 h-index	206112 48 g-index
61 all docs	61 docs citations	61 times ranked	6110 citing authors

#	Article	IF	Citations
1	Phelan-McDermid syndrome: a classification system after 30Âyears of experience. Orphanet Journal of Rare Diseases, 2022, 17, 27.	2.7	32
2	Mechanism of Disease: Recessive ADAMTSL4 Mutations and Craniosynostosis with Ectopia Lentis. Case Reports in Genetics, 2022, 2022, 1-8.	0.2	3
3	Neurodegenerative VPS41 variants inhibit HOPS function and mTORC1â€dependent TFEB/TFE3 regulation. EMBO Molecular Medicine, 2021, 13, e13258.	6.9	26
4	Changes in empowerment and anxiety of patients and parents during genetic counselling for epilepsy. European Journal of Paediatric Neurology, 2021, 32, 128-135.	1.6	4
5	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	2.4	24
6	CHARGE syndrome and related disorders: a mechanistic link. Human Molecular Genetics, 2021, 30, 2215-2224.	2.9	1
7	<scp>EPHA7</scp> haploinsufficiency is associated with a neurodevelopmental disorder. Clinical Genetics, 2021, 100, 396-404.	2.0	3
8	TAB2 deletions and variants cause a highly recognisable syndrome with mitral valve disease, cardiomyopathy, short stature and hypermobility. European Journal of Human Genetics, 2021, 29, 1669-1676.	2.8	19
9	Exome sequencing identifies the first genetic determinants of sirenomelia in humans. Human Mutation, 2020, 41, 926-933.	2.5	8
10	Under-reported aspects of diagnosis and treatment addressed in the Dutch-Flemish guideline for comprehensive diagnostics in disorders/differences of sex development. Journal of Medical Genetics, 2020, 57, 581-589.	3.2	8
11	PRRT2-related phenotypes in patients with a 16p11.2 deletion. European Journal of Medical Genetics, 2019, 62, 265-269.	1.3	20
12	Improving the diagnostic yield of exome- sequencing by predicting geneâ€“phenotype associations using large-scale gene expression analysis. Nature Communications, 2019, 10, 2837.	12.8	107
13	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
14	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	7.6	143
15	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.1	131
16	Sema3a plays a role in the pathogenesis of CHARGE syndrome. Human Molecular Genetics, 2018, 27, 1343-1352.	2.9	20
17	Imaging of Clival Hypoplasia in CHARGE Syndrome and Hypothesis for Development: A Case-Control Study. American Journal of Neuroradiology, 2018, 39, 1938-1942.	2.4	9
18	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36

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19	The phenotypic spectrum of proximal 6q deletions based on a large cohort derived from social media and literature reports. European Journal of Human Genetics, 2018, 26, 1478-1489.	2.8	31
20	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
21	Support for the Diagnosis of CHARGE Syndrome. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 634.	2.2	0
22	Developing a CHARGE syndrome checklist: Health supervision across the lifespan (from head to toe). American Journal of Medical Genetics, Part A, 2017, 173, 684-691.	1.2	32
23	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. European Journal of Human Genetics, 2017, 25, 694-701.	2.8	33
24	Copy number variation in a hospitalâ€based cohort of children with epilepsy. Epilepsia Open, 2017, 2, 244-254.	2.4	13
25	Guidelines in CHARGE syndrome and the missing link: Cranial imaging. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 450-464.	1.6	29
26	New insights and advances in CHARGE syndrome: Diagnosis, etiologies, treatments, and research discoveries. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 397-406.	1.6	46
27	Cerebellar Vermis and Midbrain Hypoplasia Upon Conditional Deletion of Chd7 from the Embryonic Mid-Hindbrain Region. Frontiers in Neuroanatomy, 2017, 11, 86.	1.7	7
28	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. Nature Genetics, 2016, 48, 877-887.	21.4	67
29	Influence of hearing loss and cognitive abilities on language development in CHARGE Syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2022-2030.	1.2	14
30	CREBBP mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2681-2693.	1.2	43
31	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. Genome Biology, 2016, 17, 243.	8.8	241
32	Congenital arch vessel anomalies in CHARGE syndrome: A frequent feature with risk for co-morbidity. IJC Heart and Vasculature, 2016, 12, 21-25.	1.1	14
33	Central Adrenal Insufficiency Is Not a Common Feature in CHARGE Syndrome: A Cross-Sectional Study in 2 Cohorts. Journal of Pediatrics, 2016, 176, 150-155.	1.8	5
34	Is there an effect of intranasal insulin on development and behaviour in Phelan-McDermid syndrome? A randomized, double-blind, placebo-controlled trial. European Journal of Human Genetics, 2016, 24, 1696-1701.	2.8	30
35	Suggestions for a Guideline for Cochlear Implantation in CHARGE Syndrome. Otology and Neurotology, 2016, 37, 1275-1283.	1.3	12
36	Duplication 2p25 in a child with clinical features of CHARGE syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1148-1154.	1.2	11

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37	Developmental phenotype in Phelan-McDermid (22q13.3 deletion) syndrome: a systematic and prospective study in 34 children. Journal of Neurodevelopmental Disorders, 2016, 8, 16.	3.1	51
38	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	2.8	1
39	Haploinsufficiency of the STX1B gene is associated with myoclonic astatic epilepsy. European Journal of Paediatric Neurology, 2016, 20, 489-492.	1.6	52
40	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	6.2	95
41	Whole-exome sequencing is a powerful approach for establishing the etiological diagnosis in patients with intellectual disability and microcephaly. BMC Medical Genomics, 2015, 9, 7.	1.5	65
42	Immune Dysfunction in Children with CHARGE Syndrome: A Cross-Sectional Study. PLoS ONE, 2015, 10, e0142350.	2.5	27
43	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	2.8	85
44	CHARGE syndrome: a review of the immunological aspects. European Journal of Human Genetics, 2015, 23, 1451-1459.	2.8	44
45	Clinical utility gene card for: CHARGE syndrome - update 2015. European Journal of Human Genetics, 2015, 23, 3-4.	2.8	18
46	Functional Insights into Chromatin Remodelling from Studies on CHARGE Syndrome. Trends in Genetics, 2015, 31, 600-611.	6.7	66
47	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. American Journal of Human Genetics, 2015, 97, 483-492.	6.2	70
48	A novel homozygous insertion and review of published mutations in the NNT gene causing familial glucocorticoid deficiency (FGD). European Journal of Medical Genetics, 2015, 58, 642-649.	1.3	24
49	Can Characteristics of Reciprocal Translocations Predict the Chance of Transferable Embryos in PGD Cycles?. Journal of Clinical Medicine, 2014, 3, 348-358.	2.4	6
50	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	2.8	109
51	CHD7 mutations are not a major cause of atrioventricular septal and conotruncal heart defects. American Journal of Medical Genetics, Part A, 2014, 164, 3003-3009.	1.2	10
52	Molecular and clinical characterization of 25 individuals with exonic deletions of NRXN1 and comprehensive review of the literature. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 388-403.	1.7	93
53	The Results ofCHD7Analysis in Clinically Well-Characterized Patients with Kallmann Syndrome. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E858-E862.	3.6	69
54	The introduction of arrays in prenatal diagnosis: A special challenge. Human Mutation, 2012, 33, 923-929.	2.5	63

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55	Mutation update on the CHD7 gene involved in CHARGE syndrome. Human Mutation, 2012, 33, 1149-1160.	2.5	224
56	Anosmia Predicts Hypogonadotropic Hypogonadism in CHARGE Syndrome. Journal of Pediatrics, 2011, 158, 474-479.	1.8	45
57	MYT1L is a candidate gene for intellectual disability in patients with 2p25.3 (2pter) deletions. American Journal of Medical Genetics, Part A, 2011, 155, 2739-2745.	1.2	53
58	A 649kb microduplication in 1p34.1, including POMGNT1, in a patient with microcephaly, coloboma and laryngomalacia; and a review of the literature. European Journal of Medical Genetics, 2009, 52, 116-119.	1.3	8
59	Exon copy number alterations of the CHD7 gene are not a major cause of CHARGE and CHARGE-like syndrome. European Journal of Medical Genetics, 2008, 51, 417-425.	1.3	33