Charlotte W Ockeloen

List of Publications by Year
in descending order

45 papers	2,013 citations	279798 23 h-index	254184 43 g-index
49 all docs	49 docs citations	49 times ranked	4801 citing authors

#	Article	IF	Citations
1	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
2	How to proceed after â€œnegativeâ€•exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. Journal of Inherited Metabolic Disease, 2022, 45, 663-681.	3.6	20
3	Missense variants in ANKRD11 cause KBG syndrome by impairment of stability or transcriptional activity of the encoded protein. Genetics in Medicine, 2022, 24, 2051-2064.	2.4	12
4	MCM complex members MCM3 and MCM7 are associated with a phenotypic spectrum from Meier-Gorlin syndrome to lipodystrophy and adrenal insufficiency. European Journal of Human Genetics, 2021, 29, 1110-1120.	2.8	16
5	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	2.8	7
6	Expanding the phenotype of <scp>ASXL3</scp>â€•related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
7	Potential added value of combined DPYD/DPD genotyping and phenotyping to prevent severe toxicity in patients with a DPYD variant and decreased dihydropyrimidine dehydrogenase enzyme activity. Journal of Oncology Pharmacy Practice, 2021, , 107815522110491.	0.9	3
8	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24
9	From man to fly â€“ convergent evidence links FBXO25 to ADHD and comorbid psychiatric phenotypes. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2020, 61, 545-555.	5.2	7
10	A novel MBD5 mutation in an intellectually disabled adult female patient with epilepsy: Suggestive of early onset dementia?. Molecular Genetics & Genomic Medicine, 2019, 7, e849.	1.2	8
11	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	1.2	23
12	The CHD8 overgrowth syndrome: A detailed evaluation of an emerging overgrowth phenotype in 27 patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 557-564.	1.6	33
13	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	32
14	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
15	Exploring the behavioral and cognitive phenotype of KBG syndrome. Genes, Brain and Behavior, 2019, 18, e12553.	2.2	12
16	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
17	Adaptive and maladaptive functioning in Kleefstra syndrome compared to other rare genetic disorders with intellectual disabilities. American Journal of Medical Genetics, Part A, 2017, 173, 1821-1830.	1.2	31
18	Identification of a de novo variant in CHUK in a patient with an EEC/AEC syndromeâ€•like phenotype and hypogammaglobulinemia. American Journal of Medical Genetics, Part A, 2017, 173, 1813-1820.	1.2	8

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19	Intellectual Profiles in KBG-Syndrome: A Wechsler Based Case-Control Study. Frontiers in Behavioral Neuroscience, 2017, 11, 248.	2.0	7
20	Identification of causative variants in TXNL4A in Burn-McKeown syndrome and isolated choanal atresia. European Journal of Human Genetics, 2017, 25, 1126-1133.	2.8	10
21	Isolated arterial calcifications of the lower extremities: A clue for NT5E mutation. International Journal of Cardiology, 2016, 212, 248-250.	1.7	12
22	Tooth agenesis and orofacial clefting: genetic brothers in arms?. Human Genetics, 2016, 135, 1299-1327.	3.8	46
23	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	2.4	58
24	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
25	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
26	Primary Cataract as a Key to Recognition of Myotonic Dystrophy Type 1. European Journal of Ophthalmology, 2015, 25, e46-e49.	1.3	9
27	Mutations in the PP2A regulatory subunit B family genesPPP2R5B,PPP2R5CandPPP2R5Dcause human overgrowth. Human Molecular Genetics, 2015, 24, 4775-4779.	2.9	85
28	Short Stature in KBG Syndrome: First Responses to Growth Hormone Treatment. Hormone Research in Paediatrics, 2015, 83, 361-364.	1.8	24
29	â€˜Splitting versus lumpingâ€™: Templeâ€“Baraitser and Zimmermannâ€“Laband Syndromes. Human Genetics, 2015, 134, 1089-1097.	3.8	24
30	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	6.2	71
31	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. European Journal of Human Genetics, 2015, 23, 1176-1185.	2.8	67
32	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	2.8	48
33	Velopharyngeal insufficiency: high detection rate of genetic abnormalities if associated with additional features. Archives of Disease in Childhood, 2014, 99, 52-57.	1.9	6
34	Variability in dentofacial phenotypes in four families with WNT10A mutations. European Journal of Human Genetics, 2014, 22, 1063-1070.	2.8	34
35	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	3.2	93
36	Quantum Metrology with a Scanning Probe Atom Interferometer. Physical Review Letters, 2013, 111, 143001.	7.8	148

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37	Prenatal diagnostic testing of the Noonan syndrome genes in fetuses with abnormal ultrasound findings. European Journal of Human Genetics, 2013, 21, 936-942.	2.8	94
38	A rare complex malformation of the hand in split hand foot malformation type 3 (SHFM3). Clinical Dysmorphology, 2013, 22, 106-108.	0.3	1
39	Duplications ofBHLHA9are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. Journal of Medical Genetics, 2012, 49, 119-125.	3.2	81
40	Congenital myopathy caused by a novel missense mutation in the CFL2 gene. Neuromuscular Disorders, 2012, 22, 632-639.	0.6	49
41	Oral application of bacterial lysate in infancy decreases the risk of atopic dermatitis in children with 1 atopic parent in a randomized, placebo-controlled trial. Journal of Allergy and Clinical Immunology, 2012, 129, 1040-1047.	2.9	89
42	Risk of colorectal and endometrial cancers in EPCAM deletion-positive Lynch syndrome: a cohort study. Lancet Oncology, The, 2011, 12, 49-55.	10.7	232
43	Two adjacent microdeletions in 8q11.2 cause a phenotype suggestive of the 22q11 deletion syndrome. Clinical Dysmorphology, 2010, 19, 137-139.	0.3	1
44	Sixteen New Cases Contributing to the Characterization of Patients with Distal 22q11.2 Microduplications. Molecular Syndromology, 2010, 1, 246-254.	0.8	31
45	CD229 (Ly9) Lymphocyte Cell Surface Receptor Interacts Homophilically through Its N-Terminal Domain and Relocalizes to the Immunological Synapse. Journal of Immunology, 2005, 174, 7033-7042.	0.8	71