Christina R Fagerberg

List of Publications by Year
in descending order

41	1,020	566801 15	476904 29
papers	citations	h-index	g-index

43	43	43	2277
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in POGLUT1, Encoding Protein O-Glucosyltransferase 1, Cause Autosomal-Dominant Dowling-Degos Disease. American Journal of Human Genetics, 2014, 94, 135-143.	2.6	136
2	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	2.6	87
3	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	1.1	80
4	Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	9.4	66
5	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	1.1	58
6	17q12 deletion and duplication syndrome in Denmarkâ€”A clinical cohort of 38 patients and review of the literature. American Journal of Medical Genetics, Part A, 2016, 170, 2934-2942.	0.7	53
7	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
8	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	2.6	46
9	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
10	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
11	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. American Journal of Human Genetics, 2019, 105, 640-657.	2.6	31
12	Heart defects and other features of the 22q11 distal deletion syndrome. European Journal of Medical Genetics, 2013, 56, 98-107.	0.7	30
13	Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. American Journal of Human Genetics, 2018, 102, 1090-1103.	2.6	29
14	Clinical characteristics and real-life diagnostic approaches in all Danish children with hereditary angioedema. Orphanet Journal of Rare Diseases, 2017, 12, 55.	1.2	25
15	Is MED13L-related intellectual disability a recognizable syndrome?. European Journal of Medical Genetics, 2019, 62, 129-136.	0.7	21
16	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	1.1	21
17	Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. Brain, 2020, 143, 94-111.	3.7	18
18	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	2.6	18

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19	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	2.6	18
20	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112.	1.4	16
21	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 880-893.	1.1	14
22	National data on the early clinical use of nonâ€invasive prenatal testing in public and private healthcare in Denmark 2013â€“2017. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 884-892.	1.3	11
23	ZTTK syndrome: Clinical and molecular findings ofÂ15 cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3740-3753.	0.7	11
24	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. Clinical Genetics, 2019, 95, 403-408.	1.0	10
25	A novel mutation affecting the arginine-137 residue of AVPR2 in dizygous twins leads to nephrogenic diabetes insipidus and attenuated urine exosome aquaporin-2. Physiological Reports, 2016, 4, e12764.	0.7	9
26	Homozygosity for SCN4A Arg1142Gln causes congenital myopathy with variable disease expression. Neurology: Genetics, 2018, 4, e267.	0.9	9
27	Mutations affecting the N-terminal domains of SHANK3 point to different pathomechanisms in neurodevelopmental disorders. Scientific Reports, 2022, 12, 902.	1.6	9
28	A novel PDGFRB sequence variant in a family with a mild form of primary familial brain calcification: a case report and a review of the literature. BMC Neurology, 2019, 19, 60.	0.8	8
29	Novel phenotype of syndromic premature ovarian insufficiency associated with <scp>TP63</scp> molecular defect. Clinical Genetics, 2020, 97, 779-784.	1.0	8
30	Total number of reads affects the accuracy of fetal fraction estimates in NIPT. Molecular Genetics & Genomic Medicine, 2021, 9, e1653.	0.6	8
31	A new 1p36.13â€“1p36.12 microdeletion syndrome characterized by learning disability, behavioral abnormalities, and ptosis. Clinical Genetics, 2020, 97, 927-932.	1.0	6
32	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	1.0	6
33	Variants in ADD1 cause intellectual disability, corpus callosum dysgenesis, and ventriculomegaly in humans. Genetics in Medicine, 2022, 24, 319-331.	1.1	6
34	Genotype-Phenotype Comparison in POGZ-Related Neurodevelopmental Disorders by Using Clinical Scoring. Genes, 2022, 13, 154.	1.0	6
35	Chromosomal Aberrations in Monozygotic and Dizygotic Twins Versus Singletons in Denmark During 1968â€“2009. Twin Research and Human Genetics, 2017, 20, 216-225.	0.3	3
36	Trisomy 8 mosaicism in the placenta: A Danish cohort study of 37 cases and a literature review. Prenatal Diagnosis, 2021, 41, 409-421.	1.1	3

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37	Phenotypic heterogeneity and mosaicism in Xia-Gibbs syndrome: Five Danish patients with novel variants in AHDC1. European Journal of Medical Genetics, 2021, 64, 104280.	0.7	3
38	Carriers of <scp>COL3A1</scp> pathogenic variants in Denmark: Interfamilial variability in severity and outcome of elective surgical procedures. Clinical Genetics, 2022, 102, 191-200.	1.0	3
39	Monoâ€allelic loss of <scp>YTHDF3</scp> and neurodevelopmental disorder: clinical features of four individuals with 8q12.3 deletions. Clinical Genetics, 2022, 101, 208-213.	1.0	2
40	Prostaglandin E₂â€<scp>EP</scp>₃ receptor subtype gene deletion in mother and son impairs platelet aggregation. British Journal of Haematology, 2019, 184, 851-853.	1.2	0
41	Transient congenital hyperinsulinism and hemolytic disease of a newborn despite rhesus D prophylaxis: a case report. Journal of Medical Case Reports, 2021, 15, 573.	0.4	0