# Christina R Fagerberg

List of Publications by Year
in descending order



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|---|---|---|---|
| 41 papers | 1,020 citations | 567281 15 h-index | 477307 29 g-index |
| 43 all docs | 43 docs citations | 43 times ranked | 2277 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. | 6.2 | 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. | 6.2 | 87 |
| 3 | The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307. | 2.4 | 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. | 21.4 | 66 |
| 5 | PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814. | 2.4 | 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. | 1.2 | 53 |
| 7 | Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63. | 8.2 | 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. | 6.2 | 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, . | 10.3 | 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. | 2.4 | 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. | 6.2 | 31 |
| 12 | Heart defects and other features of the 22q11 distal deletion syndrome. European Journal of Medical Genetics, 2013, 56, 98-107. | 1.3 | 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. | 6.2 | 29 |
| 14 | Clinical characteristics and real-life diagnostic approaches in all Danish children with hereditary angioedema. Orphanet Journal of Rare Diseases, 2017, 12, 55. | 2.7 | 25 |
| 15 | Is MED13L-related intellectual disability a recognizable syndrome?. European Journal of Medical Genetics, 2019, 62, 129-136. | 1.3 | 21 |
| 16 | Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159. | 2.4 | 21 |
| 17 | Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. Brain, 2020, 143, 94-111. | 7.6 | 18 |
| 18 | NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976. | 6.2 | 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. | 6.2 | 18 |
| 20 | Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112. | 2.8 | 16 |
| 21 | Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 880-893. | 2.4 | 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. | 2.8 | 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. | 1.2 | 11 |
| 24 | Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. Clinical Genetics, 2019, 95, 403-408. | 2.0 | 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. | 1.7 | 9 |
| 26 | Homozygosity for SCN4A Arg1142Gln causes congenital myopathy with variable disease expression. Neurology: Genetics, 2018, 4, e267. | 1.9 | 9 |
| 27 | Mutations affecting the N-terminal domains of SHANK3 point to different pathomechanisms in neurodevelopmental disorders. Scientific Reports, 2022, 12, 902. | 3.3 | 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. | 1.8 | 8 |
| 29 | Novel phenotype of syndromic premature ovarian insufficiency associated with <scp>TP63</scp> molecular defect. Clinical Genetics, 2020, 97, 779-784. | 2.0 | 8 |
| 30 | Total number of reads affects the accuracy of fetal fraction estimates in NIPT. Molecular Genetics & Genomic Medicine, 2021, 9, e1653. | 1.2 | 8 |
| 31 | A new 1p36.13â€“1p36.12 microdeletion syndrome characterized by learning disability, behavioral abnormalities, and ptosis. Clinical Genetics, 2020, 97, 927-932. | 2.0 | 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. | 2.0 | 6 |
| 33 | Variants in ADD1 cause intellectual disability, corpus callosum dysgenesis, and ventriculomegaly in humans. Genetics in Medicine, 2022, 24, 319-331. | 2.4 | 6 |
| 34 | Genotype-Phenotype Comparison in POGZ-Related Neurodevelopmental Disorders by Using Clinical Scoring. Genes, 2022, 13, 154. | 2.4 | 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. | 0.6 | 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. | 2.3 | 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. | 1.3 | 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. | 2.0 | 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. | 2.0 | 2 |
| 40 | Prostaglandin E<sub>2</sub>â€<scp>EP</scp><sub>3</sub> receptor subtype gene deletion in mother and son impairs platelet aggregation. British Journal of Haematology, 2019, 184, 851-853. | 2.5 | 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. | 0.8 | 0 |