# Christina R Fagerberg

## List of Publications by Citations



| 36 | 498 | 12 | 21 |
|---|---|---|---|
| papers | citations | h-index | g-index |
| 43 | 768 | 6.7 | 2.76 |
| ext. papers | ext. citations | avg, IF | L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 36 | Mutations in POGLUT1, encoding protein O-glucosyltransferase 1, cause autosomal-dominant Dowling-Degos disease. *American Journal of Human Genetics*, **2014**, 94, 135-43 | 11 | 110 |
| 35 | 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 | 11 | 44 |
| 34 | Biallelic mutations in the 3'-5' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. *Nature Genetics*, **2017**, 49, 457-464 | 36.3 | 43 |
| 33 | The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. *Genetics in Medicine*, **2019**, 21, 1295-1307 | 8.1 | 36 |
| 32 | 17q12 deletion and duplication syndrome in Denmark-A clinical cohort of 38 patients and review of the literature. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2934-2942 | 2.5 | 28 |
| 31 | Heart defects and other features of the 22q11 distal deletion syndrome. *European Journal of Medical Genetics*, **2013**, 56, 98-107 | 2.6 | 27 |
| 30 | PEDIA: prioritization of exome data by image analysis. *Genetics in Medicine*, **2019**, 21, 2807-2814 | 8.1 | 20 |
| 29 | De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. *American Journal of Human Genetics*, **2019**, 105, 283-301 | 11 | 20 |
| 28 | Clinical characteristics and real-life diagnostic approaches in all Danish children with hereditary angioedema. *Orphanet Journal of Rare Diseases*, **2017**, 12, 55 | 4.2 | 19 |
| 27 | 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 | 11 | 19 |
| 26 | 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 | 11 | 16 |
| 25 | Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6, | 14.3 | 12 |
| 24 | Is MED13L-related intellectual disability a recognizable syndrome?. *European Journal of Medical Genetics*, **2019**, 62, 129-136 | 2.6 | 11 |
| 23 | Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63 | 14.4 | 9 |
| 22 | Deletions and loss-of-function variants in TP63 associated with orofacial clefting. *European Journal of Human Genetics*, **2019**, 27, 1101-1112 | 5.3 | 7 |
| 21 | Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. *Brain*, **2020**, 143, 94-111 | 11.2 | 7 |
| 20 | Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. *Genetics in Medicine*, **2021**, 23, 1028-1040 | 8.1 | 7 |

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|---|---|---|---|
| 19 | 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 | 2.6 | 7 |
| 18 | Homozygosity for Arg1142Gln causes congenital myopathy with variable disease expression. *Neurology: Genetics*, **2018**, 4, e267 | 3.8 | 7 |
| 17 | Novel phenotype of syndromic premature ovarian insufficiency associated with TP63 molecular defect. *Clinical Genetics*, **2020**, 97, 779-784 | 4 | 6 |
| 16 | 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 | 11 | 5 |
| 15 | Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. *Clinical Genetics*, **2019**, 95, 403-408 | 4 | 5 |
| 14 | National data on the early clinical use of non-invasive prenatal testing in public and private healthcare in Denmark 2013-2017. *Acta Obstetricia Et Gynecologica Scandinavica*, **2021**, 100, 884-892 | 3.8 | 5 |
| 13 | 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 | 3.1 | 4 |
| 12 | NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. *American Journal of Human Genetics*, **2020**, 107, 963-976 | 11 | 4 |
| 11 | Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. *Genetics in Medicine*, **2021**, 23, 2150-2159 | 8.1 | 4 |
| 10 | Chromosomal Aberrations in Monozygotic and Dizygotic Twins Versus Singletons in Denmark During 1968-2009. *Twin Research and Human Genetics*, **2017**, 20, 216-225 | 2.2 | 3 |
| 9 | Total number of reads affects the accuracy of fetal fraction estimates in NIPT. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1653 | 2.3 | 3 |
| 8 | ZTTK syndrome: Clinical and molecular findings of□15 cases and a review of the literature. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3740-3753 | 2.5 | 3 |
| 7 | A new 1p36.13-1p36.12 microdeletion syndrome characterized by learning disability, behavioral abnormalities, and ptosis. *Clinical Genetics*, **2020**, 97, 927-932 | 4 | 2 |
| 6 | Mutations affecting the N-terminal domains of SHANK3 point to different pathomechanisms in neurodevelopmental disorders.. *Scientific Reports*, **2022**, 12, 902 | 4.9 | 1 |
| 5 | Epileptic encephalopathy caused by ARV1 deficiency: Refinement of the genotype-phenotype spectrum and functional impact on GPI-anchored proteins. *Clinical Genetics*, **2021**, 100, 607-614 | 4 | 1 |
| 4 | Trisomy 8 mosaicism in the placenta: A Danish cohort study of 37 cases and a literature review. *Prenatal Diagnosis*, **2021**, 41, 409-421 | 3.2 | 0 |
| 3 | Phenotypic heterogeneity and mosaicism in Xia-Gibbs syndrome: Five Danish patients with novel variants in AHDC1. *European Journal of Medical Genetics*, **2021**, 64, 104280 | 2.6 | 0 |
| 2 | Transient congenital hyperinsulinism and hemolytic disease of a newborn despite rhesus D prophylaxis: a case report. *Journal of Medical Case Reports*, **2021**, 15, 573 | 1.2 | |

1 Prostaglandin E -EP receptor subtype gene deletion in mother and son impairs platelet aggregation. *British Journal of Haematology*, **2019**, 184, 851-853 4·5