Christina R Fagerberg

List of Publications by Year in Descending Order

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

#	Paper	IF	Citations
36	Mutations affecting the N-terminal domains of SHANK3 point to different pathomechanisms in neurodevelopmental disorders.. *Scientific Reports*, **2022**, 12, 902	4.9	1
35	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	
34	Total number of reads affects the accuracy of fetal fraction estimates in NIPT. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1653	2.3	3
33	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
32	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
31	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
30	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
29	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
28	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
27	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. *Genetics in Medicine*, **2021**, 23, 2150-2159	8.1	4
26	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
25	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. *American Journal of Human Genetics*, **2020**, 107, 963-976	11	4
24	A new 1p36.13-1p36.12 microdeletion syndrome characterized by learning disability, behavioral abnormalities, and ptosis. *Clinical Genetics*, **2020**, 97, 927-932	4	2
23	Novel phenotype of syndromic premature ovarian insufficiency associated with TP63 molecular defect. *Clinical Genetics*, **2020**, 97, 779-784	4	6
22	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
21	Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. *Brain*, **2020**, 143, 94-111	11.2	7
20	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12

19	PEDIA: prioritization of exome data by image analysis. *Genetics in Medicine*, **2019**, 21, 2807-2814	8.1	20
18	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
17	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. *European Journal of Human Genetics*, **2019**, 27, 1101-1112	5.3	7
16	Is MED13L-related intellectual disability a recognizable syndrome?. *European Journal of Medical Genetics*, **2019**, 62, 129-136	2.6	11
15	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
14	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
13	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. *Clinical Genetics*, **2019**, 95, 403-408	4	5
12	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. *Genetics in Medicine*, **2019**, 21, 1295-1307	8.1	36
11	Prostaglandin E -EP receptor subtype gene deletion in mother and son impairs platelet aggregation. *British Journal of Haematology*, **2019**, 184, 851-853	4.5	
10	Homozygosity for Arg1142Gln causes congenital myopathy with variable disease expression. *Neurology: Genetics*, **2018**, 4, e267	3.8	7
9	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
8	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
7	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
6	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
5	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
4	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
3	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
2	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

1	Heart defects and other features of the 22q11 distal deletion syndrome. *European Journal of Medical Genetics*, **2013**, 56, 98-107	2.6	27