Maria Pettersson

List of Publications by Year
in descending order

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26 papers	721 citations	623188 14 h-index	580395 25 g-index
26 all docs	26 docs citations	26 times ranked	1597 citing authors

#	Article	IF	Citations
1	Multiâ€omics analysis reveals multiple mechanisms causing Praderâ€“Willi like syndrome in a family with a X;15 translocation. Human Mutation, 2022, 43, 1567-1575.	1.1	3
2	Lissencephaly in an epilepsy cohort: Molecular, radiological and clinical aspects. European Journal of Paediatric Neurology, 2021, 30, 71-81.	0.7	22
3	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	3.6	116
4	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	1.1	19
5	Discordant structural chromosomal aberrations in chorionic villi and amniotic fluid leading to a formation of an isochromosome 21: a case report. Molecular Cytogenetics, 2021, 14, 30.	0.4	1
6	Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. Human Genetics, 2021, 140, 775-790.	1.8	9
7	Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. Frontiers in Genetics, 2021, 12, 803683.	1.1	1
8	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	1.1	12
9	Wholeâ€genome sequencing reveals complex chromosome rearrangement disrupting <scp>NIPBL</scp> in infant with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1143-1151.	0.7	17
10	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. PLoS ONE, 2020, 15, e0228622.	1.1	6
11	Confined placental mosaicism of Duchenne muscular dystrophy: a case report. Molecular Cytogenetics, 2020, 13, 51.	0.4	2
12	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68.	3.6	88
13	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858.	1.5	36
14	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. Molecular Genetics & Genomic Medicine, 2019, 7, e549.	0.6	12
15	Targeted copy number screening highlights an intragenic deletion of WDR63 as the likely cause of human occipital encephalocele and abnormal CNS development in zebrafish. Human Mutation, 2018, 39, 495-505.	1.1	17
16	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	1.5	28
17	Flanking complex copy number variants in the same family formed through unequal crossing-over during meiosis. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2018, 812, 1-4.	0.4	6
18	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	1.1	16

#	Article	IF	Citations
19	Rare Copy Number Variants in Array-Based Comparative Genomic Hybridization in Early-Onset Skeletal Fragility. Frontiers in Endocrinology, 2018, 9, 380.	1.5	20
20	Copy Number Variants Are Enriched in Individuals With Early-Onset Obesity and Highlight Novel Pathogenic Pathways. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 3029-3039.	1.8	39
21	Further evidence for specific IFIH1 mutation as a cause of Singletonâ€“Merten syndrome with phenotypic heterogeneity. American Journal of Medical Genetics, Part A, 2017, 173, 1396-1399.	0.7	28
22	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192.	1.1	58
23	Low Copy Number of the AMY1 Locus Is Associated with Early-Onset Female Obesity in Finland. PLoS ONE, 2015, 10, e0131883.	1.1	70
24	Identification of three novelFGF16mutations in Xâ€linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. Molecular Genetics & Genomic Medicine, 2014, 2, 402-411.	0.6	17
25	Different mutations inPDE4Dassociated with developmental disorders with mirror phenotypes. Journal of Medical Genetics, 2014, 51, 45-54.	1.5	57
26	Recombinant FVIIa in children with liver disease. Thrombosis Research, 2005, 116, 185-197.	0.8	21