

Maria Pettersson

List of Publications by Year in descending order

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26
papers

721
citations

623734

14
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580821

25
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26
all docs

26
docs citations

26
times ranked

1597
citing authors

#	ARTICLE	IF	CITATIONS
1	Multi-omics analysis reveals multiple mechanisms causing Prader-Willi like syndrome in a family with a X;15 translocation. <i>Human Mutation</i> , 2022, 43, 1567-1575.	2.5	3
2	Lissencephaly in an epilepsy cohort: Molecular, radiological and clinical aspects. <i>European Journal of Paediatric Neurology</i> , 2021, 30, 71-81.	1.6	22
3	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	8.2	116
4	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. <i>Journal of Human Genetics</i> , 2021, 66, 995-1008.	2.3	19
5	Discordant structural chromosomal aberrations in chorionic villi and amniotic fluid leading to a formation of an isochromosome 21: a case report. <i>Molecular Cytogenetics</i> , 2021, 14, 30.	0.9	1
6	Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. <i>Human Genetics</i> , 2021, 140, 775-790.	3.8	9
7	Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. <i>Frontiers in Genetics</i> , 2021, 12, 803683.	2.3	1
8	Cytogenetically visible inversions are formed by multiple molecular mechanisms. <i>Human Mutation</i> , 2020, 41, 1979-1998.	2.5	12
9	Whole-genome sequencing reveals complex chromosome rearrangement disrupting <i>NIPBL</i> in infant with Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1143-1151.	1.2	17
10	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. <i>PLoS ONE</i> , 2020, 15, e0228622.	2.5	6
11	Confined placental mosaicism of Duchenne muscular dystrophy: a case report. <i>Molecular Cytogenetics</i> , 2020, 13, 51.	0.9	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. <i>Genome Medicine</i> , 2019, 11, 68.	8.2	88
13	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. <i>PLoS Genetics</i> , 2019, 15, e1007858.	3.5	36
14	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e549.	1.2	12
15	Targeted copy number screening highlights an intragenic deletion of <i>WDR63</i> as the likely cause of human occipital encephalocele and abnormal CNS development in zebrafish. <i>Human Mutation</i> , 2018, 39, 495-505.	2.5	17
16	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. <i>PLoS Genetics</i> , 2018, 14, e1007780.	3.5	28
17	Flanking complex copy number variants in the same family formed through unequal crossing-over during meiosis. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2018, 812, 1-4.	1.0	6
18	<i>Alu-Alu</i> mediated intragenic duplications in <i>IFT81</i> and <i>MATN3</i> are associated with skeletal dysplasias. <i>Human Mutation</i> , 2018, 39, 1456-1467.	2.5	16

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19	Rare Copy Number Variants in Array-Based Comparative Genomic Hybridization in Early-Onset Skeletal Fragility. <i>Frontiers in Endocrinology</i> , 2018, 9, 380.	3.5	20
20	Copy Number Variants Are Enriched in Individuals With Early-Onset Obesity and Highlight Novel Pathogenic Pathways. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 3029-3039.	3.6	39
21	Further evidence for specific <i>IFIH1</i> mutation as a cause of Singletonâ€Merten syndrome with phenotypic heterogeneity. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1396-1399.	1.2	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. <i>Human Mutation</i> , 2017, 38, 180-192.	2.5	58
23	Low Copy Number of the <i>AMY1</i> Locus Is Associated with Early-Onset Female Obesity in Finland. <i>PLoS ONE</i> , 2015, 10, e0131883.	2.5	70
24	Identification of three novel <i>FGF16</i> mutations in X-linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 402-411.	1.2	17
25	Different mutations in <i>PDE4D</i> associated with developmental disorders with mirror phenotypes. <i>Journal of Medical Genetics</i> , 2014, 51, 45-54.	3.2	57
26	Recombinant FVIIa in children with liver disease. <i>Thrombosis Research</i> , 2005, 116, 185-197.	1.7	21