

Alexej Knaus

List of Publications by Year in descending order

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Version: 2024-02-01

27
papers

695
citations

623734

14
h-index

610901

24
g-index

31
all docs

31
docs citations

31
times ranked

1483
citing authors

#	ARTICLE	IF	CITATIONS
1	Occurrence of a paroxysmal nocturnal hemoglobinuria clone in an essential thrombocythemia: a link between <i>PIGV</i> and <i>MPL</i> . <i>Haematologica</i> , 2022, 107, 1989-1993.	3.5	2
2	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. <i>Nature Genetics</i> , 2022, 54, 349-357.	21.4	73
3	<i>PIGN</i> encephalopathy: Characterizing the epileptology. <i>Epilepsia</i> , 2022, 63, 974-991.	5.1	4
4	CADA: phenotype-driven gene prioritization based on a case-enriched knowledge graph. <i>NAR Genomics and Bioinformatics</i> , 2021, 3, lqab078.	3.2	20
5	TBK1 and TNFRSF13B mutations and an autoinflammatory disease in a child with lethal COVID-19. <i>Npj Genomic Medicine</i> , 2021, 6, 55.	3.8	38
6	A CRISPR-Cas9 engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	7.1	8
7	Induction of Rosette-to-Lumen stage embryoids using reprogramming paradigms in ESCs. <i>Nature Communications</i> , 2021, 12, 7322.	12.8	6
8	Evidence of the milder phenotypic spectrum of c. 1582G >A <i>PIGT</i> variant: Delineation based on seven novel Polish patients. <i>Clinical Genetics</i> , 2020, 98, 468-476.	2.0	7
9	Lessons learned from 40 novel <i>PIGA</i> patients and a review of the literature. <i>Epilepsia</i> , 2020, 61, 1142-1155.	5.1	32
10	Adult Osteosclerotic Metaphyseal Dysplasia With Progressive Osteonecrosis of the Jaws and Abnormal Bone Resorption Pattern Due to a <i>LRRK1</i> Splice Site Mutation. <i>Journal of Bone and Mineral Research</i> , 2020, 35, 1322-1332.	2.8	18
11	A Novel Mutation in <i>PIGA</i> Associated with Multiple Congenital Anomalies-Hypotonia-Seizure Syndrome 2 (MCAHS2) in a Boy with a Combination of Severe Epilepsy and Gingival Hyperplasia. <i>Molecular Syndromology</i> , 2020, 11, 30-37.	0.8	8
12	VarFish: comprehensive DNA variant analysis for diagnostics and research. <i>Nucleic Acids Research</i> , 2020, 48, W162-W169.	14.5	39
13	Mutations in <i>PIGU</i> Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. <i>American Journal of Human Genetics</i> , 2019, 105, 395-402.	6.2	39
14	Mutations in <i>PIGB</i> Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. <i>American Journal of Human Genetics</i> , 2019, 105, 384-394.	6.2	37
15	Familial Xp11.22 microdeletion including <i>SHROOM4</i> and <i>CLCN5</i> is associated with intellectual disability, short stature, microcephaly and Dent disease: a case report. <i>BMC Medical Genomics</i> , 2019, 12, 6.	1.5	6
16	Biallelic mutations in <i>PIGP</i> cause developmental and epileptic encephalopathy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 968-973.	3.7	7
17	PEDIA: prioritization of exome data by image analysis. <i>Genetics in Medicine</i> , 2019, 21, 2807-2814.	2.4	58
18	<i>PIGT</i> -CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. <i>Genetics in Medicine</i> , 2019, 21, 2216-2223.	2.4	21

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19	Complement and inflammasome overactivation mediates paroxysmal nocturnal hemoglobinuria with autoinflammation. <i>Journal of Clinical Investigation</i> , 2019, 129, 5123-5136.	8.2	36
20	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. <i>Genome Medicine</i> , 2018, 10, 3.	8.2	67
21	Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. <i>Human Mutation</i> , 2017, 38, 1394-1401.	2.5	20
22	A Novel de novo FZD2 Mutation in a Patient with Autosomal Dominant Omodysplasia. <i>Molecular Syndromology</i> , 2017, 8, 318-324.	0.8	16
23	Rare Noncoding Mutations Extend the Mutational Spectrum in the <i>PGAP3</i> Subtype of Hyperphosphatasia with Mental Retardation Syndrome. <i>Human Mutation</i> , 2016, 37, 737-744.	2.5	46
24	A homozygous HOXD13 missense mutation causes a severe form of synpolydactyly with metacarpal to carpal transformation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 615-621.	1.2	14
25	Crowdsourced direct-to-consumer genomic analysis of a family quartet. <i>BMC Genomics</i> , 2015, 16, 910.	2.8	20
26	FGFR2 mutation in a patient without typical features of Pfeiffer syndrome – The emerging role of combined NGS and phenotype based strategies. <i>European Journal of Medical Genetics</i> , 2015, 58, 376-380.	1.3	9
27	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 763-770.	6.2	37