# Alexej Knaus

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357. | 21.4 | 73 |
| 2 | Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3. | 8.2 | 67 |
| 3 | PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814. | 2.4 | 58 |
| 4 | Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. Human Mutation, 2016, 37, 737-744. | 2.5 | 46 |
| 5 | Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. American Journal of Human Genetics, 2019, 105, 395-402. | 6.2 | 39 |
| 6 | VarFish: comprehensive DNA variant analysis for diagnostics and research. Nucleic Acids Research, 2020, 48, W162-W169. | 14.5 | 39 |
| 7 | TBK1 and TNFRSF13B mutations and an autoinflammatory disease in a child with lethal COVID-19. Npj Genomic Medicine, 2021, 6, 55. | 3.8 | 38 |
| 8 | Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770. | 6.2 | 37 |
| 9 | Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. American Journal of Human Genetics, 2019, 105, 384-394. | 6.2 | 37 |
| 10 | Complement and inflammasome overactivation mediates paroxysmal nocturnal hemoglobinuria with autoinflammation. Journal of Clinical Investigation, 2019, 129, 5123-5136. | 8.2 | 36 |
| 11 | Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155. | 5.1 | 32 |
| 12 | PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. Genetics in Medicine, 2019, 21, 2216-2223. | 2.4 | 21 |
| 13 | Crowdsourced direct-to-consumer genomic analysis of a family quartet. BMC Genomics, 2015, 16, 910. | 2.8 | 20 |
| 14 | Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. Human Mutation, 2017, 38, 1394-1401. | 2.5 | 20 |
| 15 | CADA: phenotype-driven gene prioritization based on a case-enriched knowledge graph. NAR Genomics and Bioinformatics, 2021, 3, lqab078. | 3.2 | 20 |
| 16 | Adult Osteosclerotic Metaphyseal Dysplasia With Progressive Osteonecrosis of the Jaws and Abnormal Bone Resorption Pattern Due to a LRRK1 Splice Site Mutation. Journal of Bone and Mineral Research, 2020, 35, 1322-1332. | 2.8 | 18 |
| 17 | A Novel de novo FZD2 Mutation in a Patient with Autosomal Dominant Omodysplasia. Molecular Syndromology, 2017, 8, 318-324. | 0.8 | 16 |
| 18 | A homozygous HOXD13 missense mutation causes a severe form of synpolydactyly with metacarpal to carpal transformation. American Journal of Medical Genetics, Part A, 2016, 170, 615-621. | 1.2 | 14 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | FGFR2 mutation in a patient without typical features of Pfeiffer syndrome â€“ The emerging role of combined NGS and phenotype based strategies. European Journal of Medical Genetics, 2015, 58, 376-380. | 1.3 | 9 |
| 20 | A Novel Mutation in <b><i>PIGA</i></b> Associated with Multiple Congenital Anomalies-Hypotonia-Seizure Syndrome 2 (MCAHS2) in a Boy with a Combination of Severe Epilepsy and Gingival Hyperplasia. Molecular Syndromology, 2020, 11, 30-37. | 0.8 | 8 |
| 21 | A CRISPR-Cas9â€“engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, . | 7.1 | 8 |
| 22 | Biallelic mutations in PIGP cause developmental and epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2019, 6, 968-973. | 3.7 | 7 |
| 23 | Evidence of the milder phenotypic spectrum of c. 1582G >A PIGT variant: Delineation based on seven novel Polish patients. Clinical Genetics, 2020, 98, 468-476. | 2.0 | 7 |
| 24 | Familial Xp11.22 microdeletion including SHROOM4 and CLCN5 is associated with intellectual disability, short stature, microcephaly and Dent disease: a case report. BMC Medical Genomics, 2019, 12, 6. | 1.5 | 6 |
| 25 | Induction of Rosette-to-Lumen stage embryoids using reprogramming paradigms in ESCs. Nature Communications, 2021, 12, 7322. | 12.8 | 6 |
| 26 | PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991. | 5.1 | 4 |
| 27 | Occurrence of a paroxysmal nocturnal hemoglobinuria clone in an essential thrombocythemia: a link between <i>PIGV</i> and <i>MPL</i>. Haematologica, 2022, 107, 1989-1993. | 3.5 | 2 |