Alina Kurolap

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5748398/publications.pdf

Version: 2024-02-01

33	922	687363 13	501196 28
papers	citations	h-index	g-index

37	37	37	2106
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	11.9	357
2	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2200413119.	7.1	110
3	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
4	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44
5	Loss of CD55 in Eculizumab-Responsive Protein-Losing Enteropathy. New England Journal of Medicine, 2017, 377, 87-89.	27.0	41
6	Loss of Glycine Transporter 1 Causes a Subtype of Glycine Encephalopathy with Arthrogryposis and Mildly Elevated Cerebrospinal Fluid Glycine. American Journal of Human Genetics, 2016, 99, 1172-1180.	6.2	35
7	Gaucher disease type 3c: New patients with unique presentations and review of the literature. Molecular Genetics and Metabolism, 2019, 127, 138-146.	1.1	22
8	A Unique Presentation of Infantile-Onset Colitis and Eosinophilic Disease without Recurrent Infections Resulting from a Novel Homozygous CARMIL2 Variant. Journal of Clinical Immunology, 2019, 39, 430-439.	3.8	21
9	Is one diagnosis the whole story? patients with double diagnoses. American Journal of Medical Genetics, Part A, 2016, 170, 2338-2348.	1.2	20
10	A missense mutation in ALDH1A3 causes isolated microphthalmia/anophthalmia in nine individuals from an inbred Muslim kindred. European Journal of Human Genetics, 2014, 22, 419-422.	2.8	19
11	Eculizumab Is Safe and Effective as a Longâ€term Treatment for Proteinâ€losing Enteropathy Due to CD55 Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, 325-333.	1.8	19
12	Kohlschutter-Tonz Syndrome: Clinical and Genetic Insights Gained From 16 Cases Deriving From a Close-Knit Village inÂNorthern Israel. Pediatric Neurology, 2014, 50, 421-426.	2.1	17
13	<i>LRRK2, GBA </i>and<i> SMPD1 </i>Founder Mutations and Parkinson's Disease in Ashkenazi Jews. Dementia and Geriatric Cognitive Disorders, 2016, 42, 1-6.	1.5	17
14	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
15	Establishing the role of PLVAP in protein-losing enteropathy: a homozygous missense variant leads to an attenuated phenotype. Journal of Medical Genetics, 2018, 55, 779-784.	3.2	14
16	A novel TUFM homozygous variant in a child with mitochondrial cardiomyopathy expands the phenotype of combined oxidative phosphorylation deficiency 4. Journal of Human Genetics, 2019, 64, 589-595.	2.3	14
17	Homozygosity for CHEK2 p.Gly167Arg leads to a unique cancer syndrome with multiple complex chromosomal translocations in peripheral blood karyotype. Journal of Medical Genetics, 2020, 57, 500-504.	3.2	12
18	Clinical diversity of MYH7â€related cardiomyopathies: Insights into genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2019, 179, 365-372.	1.2	10

#	Article	IF	Citations
19	BRCA1/2 mutations and FMR1 alleles are randomly distributed: a case control study. European Journal of Human Genetics, 2014, 22, 277-279.	2.8	9
20	Identification of a novel PCNT founder pathogenic variant in the Israeli Druze population. European Journal of Medical Genetics, 2020, 63, 103643.	1.3	8
21	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. American Journal of Human Genetics, 2022, 109, 518-532.	6.2	8
22	A Novel Missense Mutation in FLT4 Causes Autosomal Recessive Hereditary Lymphedema. Lymphatic Research and Biology, 2015, 13, 107-111.	1.1	6
23	Eculizumab-Responsive Adult Onset Protein Losing Enteropathy, Caused by Germline CD55-Deficiency and Complicated by Aggressive Angiosarcoma. Journal of Clinical Immunology, 2021, 41, 477-481.	3.8	6
24	A recurring NFS1 pathogenic variant causes a mitochondrial disorder with variable intra-familial patient outcomes. Molecular Genetics and Metabolism Reports, 2021, 26, 100699.	1.1	5
25	A novel heterozygous lossâ€ofâ€function DCC Netrin 1 receptor variant in prenatal agenesis of corpus callosum and review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 205-212.	1.2	4
26	Non-immune Hemolysis in Gaucher Disease and Review of the Literature. Rambam Maimonides Medical Journal, 2021, 12, e0025.	1.0	4
27	Rare Disease Diagnostics: A Single-center Experience and Lessons Learnt. Rambam Maimonides Medical Journal, 2018, 9, e0018.	1.0	4
28	â€œI Do Not Want My Baby to Suffer as I Didâ€: Prenatal and Preimplantation Genetic Diagnosis for BRCA1/2 Mutations: A Case Report and Genetic Counseling Considerations. Genetic Testing and Molecular Biomarkers, 2014, 18, 461-466.	0.7	3
29	Expertsâ€™ views on COVIDâ€19 vaccination and the impact of the pandemic on patients with Gaucher disease. British Journal of Haematology, 2021, 195, e135-e137.	2.5	3
30	CD55-deficiency in Jews of Bukharan descent is caused by the Cromer blood type Dr(aâˆ’) variant. Human Genetics, 2023, 142, 683-690.	3.8	3
31	Nonâ€immune hydrops fetalis caused by PIEZO1 compound heterozygous deletions detected only by exome sequencing. Prenatal Diagnosis, 2022, , .	2.3	3
32	A novel mutation in MYCN gene causing congenital absence of the flexor pollicis longus tendon as an unusual presentation of Feingold syndrome 1. Clinical Dysmorphology, 2021, 30, 71-75.	0.3	1
33	A novel truncating variant in the FGD1 gene associated with Aarskogâ€“Scott syndrome in a family previously diagnosed with Tel Hashomer camptodactyly. American Journal of Medical Genetics, Part A, 2021, 185, 3161-3166.	1.2	0