

Alina Kurolap

List of Publications by Year in descending order

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Version: 2024-02-01

33
papers

922
citations

777949

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h-index

563245

28
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37
all docs

37
docs citations

37
times ranked

2193
citing authors

#	ARTICLE	IF	CITATIONS
1	CD55-deficiency in Jews of Bukharan descent is caused by the Cromer blood type Dr(a ⁺) variant. <i>Human Genetics</i> , 2023, 142, 683-690.	1.8	3
2	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. <i>American Journal of Human Genetics</i> , 2022, 109, 518-532.	2.6	8
3	Non-immune hydrops fetalis caused by <i>PIEZO1</i> compound heterozygous deletions detected only by exome sequencing. <i>Prenatal Diagnosis</i> , 2022, , .	1.1	3
4	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, e2200413119.	3.3	110
5	Eculizumab-Responsive Adult Onset Protein Losing Enteropathy, Caused by Germline CD55-Deficiency and Complicated by Aggressive Angiosarcoma. <i>Journal of Clinical Immunology</i> , 2021, 41, 477-481.	2.0	6
6	A recurring NFS1 pathogenic variant causes a mitochondrial disorder with variable intra-familial patient outcomes. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100699.	0.4	5
7	Pathogenic variants in <i>SMARCA5</i> , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. <i>Science Advances</i> , 2021, 7, .	4.7	17
8	A novel truncating variant in the FGD1 gene associated with Aarskog-Scott syndrome in a family previously diagnosed with Tel Hashomer camptodactyly. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3161-3166.	0.7	0
9	Non-immune Hemolysis in Gaucher Disease and Review of the Literature. <i>Rambam Maimonides Medical Journal</i> , 2021, 12, e0025.	0.4	4
10	Experts'™ views on COVID-19 vaccination and the impact of the pandemic on patients with Gaucher disease. <i>British Journal of Haematology</i> , 2021, 195, e135-e137.	1.2	3
11	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. <i>Science Immunology</i> , 2021, 6, .	5.6	357
12	A novel mutation in MYCN gene causing congenital absence of the flexor pollicis longus tendon as an unusual presentation of Feingold syndrome 1. <i>Clinical Dysmorphology</i> , 2021, 30, 71-75.	0.1	1
13	Identification of a novel PCNT founder pathogenic variant in the Israeli Druze population. <i>European Journal of Medical Genetics</i> , 2020, 63, 103643.	0.7	8
14	Homozygosity for CHEK2 p.Gly167Arg leads to a unique cancer syndrome with multiple complex chromosomal translocations in peripheral blood karyotype. <i>Journal of Medical Genetics</i> , 2020, 57, 500-504.	1.5	12
15	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. <i>Genetics in Medicine</i> , 2020, 22, 389-397.	1.1	53
16	A novel heterozygous loss-of-function <i>DCC</i> Netrin 1 receptor variant in prenatal agenesis of corpus callosum and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 205-212.	0.7	4
17	Gaucher disease type 3c: New patients with unique presentations and review of the literature. <i>Molecular Genetics and Metabolism</i> , 2019, 127, 138-146.	0.5	22
18	A Unique Presentation of Infantile-Onset Colitis and Eosinophilic Disease without Recurrent Infections Resulting from a Novel Homozygous CARMIL2 Variant. <i>Journal of Clinical Immunology</i> , 2019, 39, 430-439.	2.0	21

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19	A novel TUFM homozygous variant in a child with mitochondrial cardiomyopathy expands the phenotype of combined oxidative phosphorylation deficiency 4. <i>Journal of Human Genetics</i> , 2019, 64, 589-595.	1.1	14
20	Eculizumab Is Safe and Effective as a Long-term Treatment for Protein-losing Enteropathy Due to CD55 Deficiency. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2019, 68, 325-333.	0.9	19
21	Clinical diversity of MYH7-related cardiomyopathies: Insights into genotype-phenotype correlations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 365-372.	0.7	10
22	Pathogenic variants in glutamyl-tRNA _{Gln} amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. <i>Nature Communications</i> , 2018, 9, 4065.	5.8	44
23	Establishing the role of PLVAP in protein-losing enteropathy: a homozygous missense variant leads to an attenuated phenotype. <i>Journal of Medical Genetics</i> , 2018, 55, 779-784.	1.5	14
24	Rare Disease Diagnostics: A Single-center Experience and Lessons Learnt. <i>Rambam Maimonides Medical Journal</i> , 2018, 9, e0018.	0.4	4
25	Loss of CD55 in Eculizumab-Responsive Protein-Losing Enteropathy. <i>New England Journal of Medicine</i> , 2017, 377, 87-89.	13.9	41
26	Is one diagnosis the whole story? patients with double diagnoses. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2338-2348.	0.7	20
27	LRRK2, GBA and SMPD1 Founder Mutations and Parkinson's Disease in Ashkenazi Jews. <i>Dementia and Geriatric Cognitive Disorders</i> , 2016, 42, 1-6.	0.7	17
28	Loss of Glycine Transporter 1 Causes a Subtype of Glycine Encephalopathy with Arthrogryposis and Mildly Elevated Cerebrospinal Fluid Glycine. <i>American Journal of Human Genetics</i> , 2016, 99, 1172-1180.	2.6	35
29	A Novel Missense Mutation in FLT4 Causes Autosomal Recessive Hereditary Lymphedema. <i>Lymphatic Research and Biology</i> , 2015, 13, 107-111.	0.5	6
30	"I Do Not Want My Baby to Suffer as I Did": Prenatal and Preimplantation Genetic Diagnosis for BRCA1/2 Mutations: A Case Report and Genetic Counseling Considerations. <i>Genetic Testing and Molecular Biomarkers</i> , 2014, 18, 461-466.	0.3	3
31	A missense mutation in ALDH1A3 causes isolated microphthalmia/anophthalmia in nine individuals from an inbred Muslim kindred. <i>European Journal of Human Genetics</i> , 2014, 22, 419-422.	1.4	19
32	BRCA1/2 mutations and FMR1 alleles are randomly distributed: a case control study. <i>European Journal of Human Genetics</i> , 2014, 22, 277-279.	1.4	9
33	Kohlschutter-Tonz Syndrome: Clinical and Genetic Insights Gained From 16 Cases Deriving From a Close-Knit Village in Northern Israel. <i>Pediatric Neurology</i> , 2014, 50, 421-426.	1.0	17