Kosuke Izumi

List of Publications by Year
in descending order

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		394421	395702
68	1,491	19	33
papers	citations	h-index	g-index

68	68	68	2788
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
2	Inborn error of metabolism patients after liver transplantation: Outcomes of 35 patients over 27â€‰years in one pediatric quaternary hospital. American Journal of Medical Genetics, Part A, 2022, 188, 1443-1447.	1.2	2
3	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	2.8	7
4	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5
5	Molecular Mechanisms Contributing to the Etiology of Congenital Diaphragmatic Hernia: A Review and Novel Cases. Journal of Pediatrics, 2022, 246, 251-265.e2.	1.8	4
6	eP170: When cfDNA screening deceives: A rare case of mosaicism for 46,XX/47,XXY with uniparental isodisomy and genital atypia. Genetics in Medicine, 2022, 24, S103-S104.	2.4	0
7	<scp>MYH7</scp> variants cause complex congenital heart disease. American Journal of Medical Genetics, Part A, 2022, 188, 2772-2776.	1.2	7
8	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
9	Mosaic <scp>RAI1</scp> variant in a <scp>Smithâ€“Magenis</scp> syndrome patient with total anomalous pulmonary venous return. American Journal of Medical Genetics, Part A, 2022, 188, 3130-3134.	1.2	1
10	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
11	Variants in <scp>NAA15</scp> cause pediatric hypertrophic cardiomyopathy. American Journal of Medical Genetics, Part A, 2021, 185, 228-233.	1.2	10
12	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
13	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. American Journal of Human Genetics, 2021, 108, 1342-1349.	6.2	9
14	Clinical utility of exome sequencing in infantile heart failure. Genetics in Medicine, 2020, 22, 423-426.	2.4	17
15	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€“phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
16	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
17	Hyperinsulinism of Kabuki Syndrome: Clinical Characteristics and Treatments. Journal of Pediatric Nursing, 2020, 52, 107.	1.5	0
18	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35

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19	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
20	Clinical and molecular spectrum of CHOPS syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1126-1138.	1.2	20
21	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610.	6.2	32
22	Cathepsin L-deficiency enhances liver regeneration after partial hepatectomy. Life Sciences, 2019, 221, 293-300.	4.3	6
23	Increased Clinical Sensitivity and Specificity of Plasma Protein N-Glycan Profiling for Diagnosing Congenital Disorders of Glycosylation by Use of Flow Injectionâ€“Electrospray Ionizationâ€“Quadrupole Time-of-Flight Mass Spectrometry. Clinical Chemistry, 2019, 65, 653-663.	3.2	40
24	Interstitial 4q Deletion Syndrome Including <i>NR3C2</i> Causing Pseudohypoaldosteronism. Molecular Syndromology, 2019, 10, 327-331.	0.8	6
25	De novo variants in Myelin regulatory factor (MYRF) as candidates of a new syndrome of cardiac and urogenital anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 969-972.	1.2	39
26	DOCK3â€related neurodevelopmental syndrome: Biallelic intragenic deletion of DOCK3 in a boy with developmental delay and hypotonia. American Journal of Medical Genetics, Part A, 2018, 176, 241-245.	1.2	14
27	Prenatal profile of Pallisterâ€Killian syndrome: Retrospective analysis of 114 pregnancies, literature review and approach to prenatal diagnosis. American Journal of Medical Genetics, Part A, 2018, 176, 2575-2586.	1.2	21
28	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
29	Variable Clinical Manifestations of Xiaâ€Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. American Journal of Medical Genetics, Part A, 2018, 176, 1890-1896.	1.2	20
30	PCDH19 -related epilepsy in a male with Klinefelter syndrome: Additional evidence supporting PCDH19 cellular interference disease mechanism. Epilepsy Research, 2018, 145, 89-92.	1.6	20
31	Cardiac Fibroma with Ventricular Tachycardia: An Unusual Clinical Presentation of Nevoid Basal Cell Carcinoma Syndrome. Molecular Syndromology, 2018, 9, 219-223.	0.8	8
32	Disorders of Transcriptional Regulation: An Emerging Category of Multiple Malformation Syndromes. Molecular Syndromology, 2016, 7, 262-273.	0.8	39
33	Mosaic ratio quantification of isochromosome 12p in Pallisterâ€“Killian syndrome using droplet digital <scp>PCR</scp>. Molecular Genetics & Genomic Medicine, 2016, 4, 257-261.	1.2	14
34	ARCN1 Mutations Cause a Recognizable Craniofacial Syndrome Due to COPI-Mediated Transport Defects. American Journal of Human Genetics, 2016, 99, 451-459.	6.2	65
35	Exome sequencingâ€based identification of mutations in nonâ€syndromic genes among individuals with apparently syndromic features. American Journal of Medical Genetics, Part A, 2016, 170, 2889-2894.	1.2	26
36	Fetal akinesia deformation sequence due to a congenital disorder of glycosylation. American Journal of Medical Genetics, Part A, 2015, 167, 2411-2417.	1.2	12

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37	Exome Sequencing Identification of <i>EP300</i> Mutation in a Proband with Coloboma and Imperforate Anus: Possible Expansion of the Phenotypic Spectrum of Rubinstein-Taybi Syndrome. Molecular Syndromology, 2015, 6, 99-103.	0.8	10
38	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. Nature Genetics, 2015, 47, 338-344.	21.4	109
39	The Deubiquitinating Enzyme USP7 Regulates Androgen Receptor Activity by Modulating Its Binding to Chromatin. Journal of Biological Chemistry, 2015, 290, 21713-21723.	3.4	50
40	Pallisterâ€“Killian syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 406-413.	1.6	51
41	Cardiac manifestations of Pallisterâ€“Killian syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1130-1135.	1.2	19
42	NKX2.5 mutation identification on exome sequencing in a patient with heterotaxy. European Journal of Medical Genetics, 2014, 57, 558-561.	1.3	13
43	12p microRNA expression in fibroblast cell lines from probands with Pallister-Killian syndrome. Chromosome Research, 2014, 22, 453-461.	2.2	12
44	Genome-Wide Expression Analysis in Fibroblast Cell Lines from Probands with Pallister Killian Syndrome. PLoS ONE, 2014, 9, e108853.	2.5	14
45	Mosaic maternal uniparental disomy of chromosome 15 in Praderâ€“Willi syndrome: Utility of genomeâ€wide SNP array. American Journal of Medical Genetics, Part A, 2013, 161, 166-171.	1.2	17
46	Congenital heart defects in oculodentodigital dysplasia: Report of two cases. American Journal of Medical Genetics, Part A, 2013, 161, 3150-3154.	1.2	19
47	Endocrine phenotype of 6q16.1â€“q21 deletion involving SIM1 and Praderâ€“Willi syndromeâ€like features. American Journal of Medical Genetics, Part A, 2013, 161, 3137-3143.	1.2	36
48	Novel <scp>MBTPS</scp>2 Missense Mutation in the Nâ€Terminus Transmembrane Domain in a Patient with Ichthyosis Follicularis, Alopecia, and Photophobia Syndrome. Pediatric Dermatology, 2013, 30, e263-4.	0.9	5
49	Novel clinical manifestations in Pallisterâ€“Killian syndrome: Comprehensive evaluation of 59 affected individuals and review of previously reported cases. American Journal of Medical Genetics, Part A, 2012, 158A, 3002-3017.	1.2	80
50	Duplication 12p and Pallisterâ€“Killian syndrome: A case report and review of the literature toward defining a Pallisterâ€“Killian syndrome minimal critical region. American Journal of Medical Genetics, Part A, 2012, 158A, 3033-3045.	1.2	40
51	Utility of SNP arrays in detecting, quantifying, and determining meiotic origin of tetrasomy 12p in blood from individuals with Pallisterâ€“Killian syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 3046-3053.	1.2	41
52	1.9â€‰Mb microdeletion of 21q22.11 within Braddockâ€“Carey contiguous gene deletion syndrome region: Dissecting the phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 1535-1541.	1.2	19
53	Underlying Genetic Diagnosis of Pierre Robin Sequence: Retrospective Chart Review at Two Childrenâ€™s Hospitals and a Systematic Literature Review. Journal of Pediatrics, 2012, 160, 645-650.e2.	1.8	126
54	Familial 9q22.3 microduplication spanning PTCH1 causes short stature syndrome with mild intellectual disability and dysmorphic features. American Journal of Medical Genetics, Part A, 2011, 155, 1384-1389.	1.2	14

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55	8p21 microdeletion in a patient with intellectual disability and behavioral abnormalities. American Journal of Medical Genetics, Part A, 2011, 155, 3148-3152.	1.2	2
56	Fabrication of robust PbLa(Zr,Ti)O3 capacitor structures using insulating oxide encapsulation layers for FeRAM integration. Electronics Letters, 2011, 47, 486.	1.0	2
57	Late manifestations of trichoâ€rhinoâ€pharangeal syndrome in a patient: Expanded skeletal phenotype in adulthood. American Journal of Medical Genetics, Part A, 2010, 152A, 2115-2119.	1.2	13
58	Submicroscopic familial chromosomal translocation between 7q and 12p mimicking an autosomal dominant holoprosencephaly syndrome. Clinical Genetics, 2010, 78, 402-404.	2.0	2
59	Diaphragm dysfunction with congenital cytomegalovirus infection. Journal of Perinatology, 2010, 30, 691-694.	2.0	5
60	Caudal Regression and Tracheoesophageal Malformation Induced by Adriamycin: A Novel Chick Model of VATER Association. Pediatric Research, 2009, 65, 607-612.	2.3	9
61	Hepatitis C Virus Impairs p53 via Persistent Overexpression of 3Î²-Hydroxysterol Î”24-Reductase. Journal of Biological Chemistry, 2009, 284, 36442-36452.	3.4	58
62	Tietz syndrome: unique phenotype specific to mutations of MITF nuclear localization signal. Clinical Genetics, 2008, 74, 93-95.	2.0	28
63	Identification of a Prosencephalic-Specific Enhancer of SALL1: Comparative Genomic Approach Using the Chick Embryo. Pediatric Research, 2007, 61, 660-665.	2.3	10
64	Multiplex PCR/Liquid Chromatography Assay for Screening of Subtelomeric Rearrangements. Genetic Testing and Molecular Biomarkers, 2007, 11, 241-248.	1.7	4
65	Screening for Alagille Syndrome Mutations in the JAG1 and NOTCH2 Genes Using Denaturing High-Performance Liquid Chromatography. Genetic Testing and Molecular Biomarkers, 2007, 11, 216-227.	1.7	7
66	Partial Deletion of LIS1: A Pitfall in Molecular Diagnosis of Miller-Dieker Syndrome. Pediatric Neurology, 2007, 36, 258-260.	2.1	8
67	EFNB1 mutation at the ephrin ligand-receptor dimerization interface in a patient with craniofrontonasal syndrome. Congenital Anomalies (discontinued), 2007, 47, 49-52.	0.6	15
68	Screening for Partial Deletions in the CREBBP Gene in Rubinsteinâ€“Taybi Syndrome Patients Using Multiplex PCR/Liquid Chromatography. Genetic Testing and Molecular Biomarkers, 2006, 10, 265-271.	1.7	12