Ken Saida

List of Publications by Year
in descending order

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42 papers	501 citations	687363 13 h-index	752698 20 g-index
44 all docs	44 docs citations	44 times ranked	1044 citing authors

#	Article	IF	Citations
1	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	3.2	4
2	Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function GRIA3 variant. Human Genetics, 2022, 141, 283-293.	3.8	6
3	Monogenic causes of pigmentary mosaicism. Human Genetics, 2022, , .	3.8	2
4	Clinical course of a Japanese patient with developmental delay linked to a small 6q16.1 deletion. Human Genome Variation, 2022, 9, 14.	0.7	2
5	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
6	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	3.7	4
7	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	2.3	4
8	Pathogenic variants in the survival of motor neurons complex gene <scp>GEMIN5</scp> cause cerebellar atrophy. Clinical Genetics, 2021, 100, 722-730.	2.0	15
9	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. Science Advances, 2021, 7, .	10.3	25
10	Father-to-offspring transmission of extremely long NOTCH2NLC repeat expansions with contractions: genetic and epigenetic profiling with long-read sequencing. Clinical Epigenetics, 2021, 13, 204.	4.1	22
11	A novel truncating PAX2 mutation in a boy with renal coloboma syndrome with focal segmental glomerulosclerosis causing rapid progression to end-stage kidney disease. CEN Case Reports, 2020, 9, 19-23.	0.9	8
12	Autosomal dominant Alport syndrome due to a COL4A4 mutation with an additional ESPN variant detected by whole-exome analysis. CEN Case Reports, 2020, 9, 59-64.	0.9	0
13	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599.	2.5	6
14	Influenza virus vaccination in children with nephrotic syndrome: insignificant risk of relapse. Clinical and Experimental Nephrology, 2020, 24, 1069-1076.	1.6	9
15	A simple, refined approach to diagnosing renovascular hypertension in children: A 10â€year study. Pediatrics International, 2020, 62, 937-943.	0.5	6
16	Childhood Nephrotic Syndrome Complicated by Catastrophic Multiple Arterial Thrombosis Requiring Bilateral Above-Knee Amputation. Frontiers in Pediatrics, 2020, 8, 107.	1.9	5
17	Bardetâ€“Biedl syndrome and related disorders in Japan. Journal of Human Genetics, 2020, 65, 847-853.	2.3	9
18	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. Journal of Human Genetics, 2019, 64, 885-890.	2.3	11

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19	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
20	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. Nature Communications, 2019, 10, 2506.	12.8	46
21	Pharmacokinetics and Pharmacodynamics Estimation of Eculizumab in a 2-Year-Old Girl With Atypical Hemolytic Uremic Syndrome: A Case Report With 4-Year Follow-Up. Frontiers in Pediatrics, 2019, 7, 519.	1.9	3
22	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	1.2	10
23	Periventricular small cystic lesions in a patient with Coffin-Lowry syndrome who exhibited a novel mutation in the RPS6KA3 gene. Brain and Development, 2018, 40, 566-569.	1.1	4
24	Coagulopathy as a complication of kidney biopsies in paediatric systemic lupus erythematosus patients with antiphospholipid syndrome. Nephrology, 2018, 23, 592-596.	1.6	2
25	Treatment of hemolytic uremic syndrome related to Bordetella pertussis infection â€"is plasma exchange or eculizumab use necessary?. BMC Nephrology, 2018, 19, 365.	1.8	5
26	Azathioprine-induced Agranulocytosis and Severe Alopecia After Kidney Transplantation Associated With a NUDT15 Polymorphism: AÂCase Report. Transplantation Proceedings, 2018, 50, 3925-3927.	0.6	6
27	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
28	Diversity of renal phenotypes in patients with WDR19 mutations: Two case reports. Nephrology, 2017, 22, 566-571.	1.6	12
29	Successful therapy switch from eculizumab to mycophenolate mofetil in a girl with DEAP-HUS. Pediatric Nephrology, 2017, 32, 1997-1998.	1.7	3
30	Ampicillinâ€•and ampicillin/sulbactamâ€•resistant <scp>Escherichia coli</scp> infection in a neonatal intensive care unit in Japan. Pediatrics International, 2016, 58, 537-539.	0.5	2
31	Three cases of C3 glomerulonephritis associated with group A streptococcus infection. Immunobiology, 2016, 221, 1161.	1.9	0
32	Efficacy and safety of eculizumab in childhood atypical hemolytic uremic syndrome in Japan. Clinical and Experimental Nephrology, 2016, 20, 265-272.	1.6	22
33	The First Nationwide Survey and Genetic Analyses of Bardet-Biedl Syndrome in Japan. PLoS ONE, 2015, 10, e0136317.	2.5	16
34	Pancreatic desmoid tumor in a 4-year-old male with hemihypertrophy. Journal of Pediatric Surgery Case Reports, 2015, 3, 344-347.	0.2	1
35	Exfoliative toxin A staphylococcal scalded skin syndrome in preterm infants. European Journal of Pediatrics, 2015, 174, 551-555.	2.7	21
36	Rituximab-associated agranulocytosis in children with refractory idiopathic nephrotic syndrome: case series and review of literature. Nephrology Dialysis Transplantation, 2015, 30, 91-96.	0.7	45

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37	A case of adult Dent disease in Japan with advanced chronic kidney disease. CEN Case Reports, 2014, 3, 132-138.	0.9	4
38	A case of Bardet-Biedl syndrome complicated with intracranial hypertension in a Japanese child. Brain and Development, 2014, 36, 721-724.	1.1	10
39	Preoperative left ventricular internal dimension in end-diastole as earlier identification of early patent ductus arteriosus operation and postoperative intensive care in very low birth weight infants. Early Human Development, 2013, 89, 821-823.	1.8	6
40	Lamellar ichthyosis in a collodion baby caused by CYP4F22 mutations in a non-consanguineous family outside the Mediterranean. Journal of Dermatological Science, 2013, 72, 193-195.	1.9	23
41	Right-to-left shunting in the ductus arteriosus is induced readily by intense crying and rapid postural change in neonates with meconium-stained amniotic fluid*. Pediatric Critical Care Medicine, 2012, 13, 60-65.	0.5	4
42	Successful Unrelated Cord Blood Transplantation Using a Reduced-Intensity Conditioning Regimen in a 6-Month-Old Infant with Congenital Neutropenia Complicated by Severe Pneumonia. International Journal of Hematology, 2004, 80, 287-290.	1.6	14