

Yuri A Zarate

List of Publications by Year in descending order

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Version: 2024-02-01

83
papers

1,897
citations

394421

19
h-index

302126

39
g-index

85
all docs

85
docs citations

85
times ranked

3411
citing authors

#	ARTICLE	IF	CITATIONS
1	Fabry's disease. <i>Lancet</i> , The, 2008, 372, 1427-1435.	13.7	628
2	<i>SATB2</i> -associated syndrome: Mechanisms, phenotype, and practical recommendations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 327-337.	1.2	81
3	A dyadic approach to the delineation of diagnostic entities in clinical genomics. <i>American Journal of Human Genetics</i> , 2021, 108, 8-15.	6.2	71
4	Redefining the Etiologic Landscape of Cerebellar Malformations. <i>American Journal of Human Genetics</i> , 2019, 105, 606-615.	6.2	61
5	Natural history and genotype-phenotype correlations in 72 individuals with <i>SATB2</i> -associated syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 925-935.	1.2	57
6	Experience with hemihyperplasia and Beckwith-Wiedemann syndrome surveillance protocol. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1691-1697.	1.2	47
7	Further supporting evidence for the <i>SATB2</i> -associated syndrome found through whole exome sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1026-1032.	1.2	42
8	Genetic Causes of Macroglossia: Diagnostic Approach. <i>Pediatrics</i> , 2012, 129, e431-e437.	2.1	38
9	Mutation update for the <i>SATB2</i> gene. <i>Human Mutation</i> , 2019, 40, 1013-1029.	2.5	38
10	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. <i>Brain</i> , 2020, 143, 55-68.	7.6	38
11	Genotype and phenotype in 12 additional individuals with <i>SATB2</i> -associated syndrome. <i>Clinical Genetics</i> , 2017, 92, 423-429.	2.0	33
12	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	6.2	30
13	De novo variants in EBF3 are associated with hypotonia, developmental delay, intellectual disability, and autism. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a002097.	1.2	28
14	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. <i>Genetics in Medicine</i> , 2021, 23, 363-373.	2.4	28
15	Lessons from a pair of siblings with BPAN. <i>European Journal of Human Genetics</i> , 2016, 24, 1080-1083.	2.8	26
16	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1338-1347.	2.4	25
17	Evaluation of Growth in Patients With Isolated Cleft Lip and/or Cleft Palate. <i>Pediatrics</i> , 2010, 125, e543-e549.	2.1	22
18	Infrequent Manifestations of Kabuki Syndrome in a Patient with Novel <i>MLL2</i> Mutation. <i>Molecular Syndromology</i> , 2012, 3, 180-184.	0.8	22

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19	Cardiovascular and genitourinary anomalies in patients with duplications within the Williams syndrome critical region: Phenotypic expansion and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1998-2002.	1.2	22
20	Frequency and age at occurrence of clinical manifestations of disease in patients with hypophosphatasia: a systematic literature review. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 85.	2.7	21
21	Activating variants in <i>PDGFRB</i> result in a spectrum of disorders responsive to imatinib monotherapy. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1576-1591.	1.2	21
22	A new case of de novo 11q duplication in a patient with normal development and intelligence and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 265-270.	1.2	20
23	De novo variants in <i>SNAP25</i> cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021, 23, 653-660.	2.4	20
24	Unique Cerebrovascular Anomalies in Noonan Syndrome With <i>RAF1</i> Mutation. <i>Journal of Child Neurology</i> , 2014, 29, NP13-NP17.	1.4	19
25	<i>COL1A1</i> and <i>COL1A2</i> sequencing results in cohort of patients undergoing evaluation for potential child abuse. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1858-1862.	1.2	19
26	Variants in the degron of <i>AFF3</i> are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 857-873.	6.2	19
27	<i>SMARCE1</i> , a rare cause of Coffin-Siris Syndrome: Clinical description of three additional cases. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1967-1973.	1.2	18
28	Bone health and <i>SATB2</i> associated syndrome. <i>Clinical Genetics</i> , 2018, 93, 588-594.	2.0	18
29	Constitutive activation of the PI3K-AKT pathway and cardiovascular abnormalities in an individual with Kosaki overgrowth syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1047-1052.	1.2	18
30	Novel Fibrillin 1 Mutation in a Case of Neonatal Marfan Syndrome: The Increasing Importance of Early Recognition. <i>Congenital Heart Disease</i> , 2007, 2, 342-346.	0.2	17
31	Lethal presentation of neurofibromatosis and Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1360-1366.	1.2	17
32	Papillary thyroid cancer in a patient with interstitial 6q25 deletion including <i>ARID1B</i> . <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1857-1859.	1.2	17
33	Description of the First Case of Adenomyomatosis of the Gallbladder in an Infant. <i>Case Reports in Pediatrics</i> , 2014, 2014, 1-3.	0.4	16
34	<i>HIST1H1E</i> heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the <i>HIST1H1E</i> syndrome phenotype in 30 individuals. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2049-2055.	1.2	16
35	Using facial analysis technology in a typical genetic clinic: experience from 30 individuals from a single institution. <i>Journal of Human Genetics</i> , 2019, 64, 1243-1245.	2.3	16
36	The role of <i>IQSEC2</i> in syndromic intellectual disability: Narrowing the diagnostic odyssey. , 2017, 173, 2814-2820.		14

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37	Satb2 regulates proliferation and nuclear integrity of pre-osteoblasts. <i>Bone</i> , 2019, 127, 488-498.	2.9	14
38	STAR syndrome is part of the differential diagnosis of females with anorectal malformations. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1940-1943.	1.2	13
39	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021, 23, 374-383.	2.4	13
40	Growth, development, and phenotypic spectrum of individuals with deletions of 2q33.1 involving <i>SATB2</i> . <i>Clinical Genetics</i> , 2021, 99, 547-557.	2.0	13
41	Nuclear radiation and prevalence of structural birth defects among infants born to women from the Marshall Islands. <i>Birth Defects Research</i> , 2019, 111, 1192-1204.	1.5	11
42	A case of minimal change disease in a Fabry patient. <i>Pediatric Nephrology</i> , 2010, 25, 553-556.	1.7	10
43	Severe neonatal presentation of Kleeftstra syndrome in a patient with hypoplastic left heart syndrome and 9q34.3 microdeletion. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2014, 100, 985-990.	1.6	10
44	First clinical report of an infant with microcephaly and <i>CASC5</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2215-2218.	1.2	10
45	Additional <i>de novo</i> missense genetic variants in <i>NALCN</i> associated with <i>CLIFAHDD</i> syndrome. <i>Clinical Genetics</i> , 2017, 91, 929-931.	2.0	10
46	Speech, language, and feeding phenotypes of <i>SATB2</i> -associated syndrome. <i>Clinical Genetics</i> , 2019, 96, 485-492.	2.0	10
47	<i>SATB2</i> -associated syndrome (SAS) and associated dental findings. <i>Special Care in Dentistry</i> , 2019, 39, 220-224.	0.8	10
48	Epilepsy and Electroencephalographic Abnormalities in <i>SATB2</i> -Associated Syndrome. <i>Pediatric Neurology</i> , 2020, 112, 94-100.	2.1	10
49	Aortic dilation in pediatric patients. <i>European Journal of Pediatrics</i> , 2015, 174, 1585-1592.	2.7	9
50	Ophthalmic findings in patients with arterial tortuosity syndrome and carriers: A case series. <i>Ophthalmic Genetics</i> , 2018, 39, 29-34.	1.2	9
51	Widening phenotypic spectrum of <i>GABBR2</i> mutation. <i>Acta Neurologica Belgica</i> , 2019, 119, 493-496.	1.1	8
52	Behavioral phenotype and sleep problems in <i>SATB2</i> -associated syndrome. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 827-832.	2.1	8
53	Advanced cardiovascular imaging in Williams syndrome: Abnormalities, usefulness, and strategy for use. <i>Journal of Magnetic Resonance Imaging</i> , 2017, 173, 1194-1199.		7
54	Dental radiographic findings in 18 individuals with <i>SATB2</i> -associated syndrome. <i>Clinical Oral Investigations</i> , 2018, 22, 2947-2951.	3.0	7

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55	CDK19-related disorder results from both loss-of-function and gain-of-function de novo missense variants. <i>Genetics in Medicine</i> , 2021, 23, 1050-1057.	2.4	7
56	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. <i>European Journal of Medical Genetics</i> , 2020, 63, 103817.	1.3	6
57	Persistent congenital hyperinsulinism in two patients with Beckwith-Wiedemann syndrome due to mosaic uniparental disomy 11p. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014, 27, 951-5.	0.9	5
58	47, XY, +der(Y),t(X;Y)(p21.1;p11.2): A unique case of XY sex reversal. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 386-391.	1.2	4
59	Description of another case of 3q26.33-3q27.2 microdeletion supports a recognizable phenotype. <i>European Journal of Medical Genetics</i> , 2013, 56, 624-625.	1.3	4
60	Aortic dilation, genetic testing, and associated diagnoses. <i>Genetics in Medicine</i> , 2016, 18, 356-363.	2.4	4
61	Severe Metabolic Acidosis and Hepatopathy due to Leukoencephalopathy with Thalamus and Brainstem Involvement and High Lactate. <i>Neuropediatrics</i> , 2017, 48, 108-110.	0.6	4
62	Individuals with SATB2-associated syndrome with and without autism have a recognizable metabolic profile and distinctive cellular energy metabolism alterations. <i>Metabolic Brain Disease</i> , 2021, 36, 1049-1056.	2.9	4
63	SATB2-associated syndrome in adolescents and adults. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2391-2398.	1.2	4
64	Intestinal Malrotation in a Patient with Pfeiffer Syndrome Type 2. <i>Cleft Palate-Craniofacial Journal</i> , 2010, 47, 638-641.	0.9	3
65	Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2168-2175.	1.2	3
66	An infant with ash-leaf and café au lait spots: a case of double phakomatosis. <i>Acta Neurologica Belgica</i> , 2017, 117, 323-324.	1.1	3
67	Less common underlying genetic diagnoses found in a cohort of 139 individuals surgically corrected for craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 487-491.	1.2	3
68	Craniosynostosis is a feature of Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1280-1286.	1.2	3
69	Growth in individuals with SATB2-associated syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2952-2957.	1.2	3
70	Twin-twin transfusion resulting in fetal cell contamination in Beckwith-Wiedemann syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1569-1570.	1.2	2
71	Genetic and phenotypic heterogeneity in KIAA0753-related ciliopathies. <i>American Journal of Medical Genetics, Part A</i> , 2021, . .	1.2	2
72	The landscape of submicroscopic structural variants at the OPN1LW/OPN1MW gene cluster on Xq28 underlying blue cone monochromacy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	7.1	2

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73	Phenotypic and microscopic description of a new case of Ermine phenotype. American Journal of Medical Genetics, Part A, 2009, 149A, 1253-1256.	1.2	1
74	Managing Sleep and Behavioral Problems in a Preschooler with SATB2-Associated Syndrome. Case Reports in Genetics, 2020, 2020, 1-4.	0.2	1
75	A clinical scoring system for early onset (neonatal) Marfan syndrome. Genetics in Medicine, 2022, , .	2.4	1
76	Prevalence and Outcomes of Primary Left Ventricular Dysfunction in Marfan Syndrome. American Journal of Cardiology, 2022, 175, 119-126.	1.6	1
77	Clinical utility of the Xâ€chromosome array. American Journal of Medical Genetics, Part A, 2013, 161, 120-130.	1.2	0
78	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
79	Aortic Geometry in Patients with Duplication 7q11.23 Compared to Healthy Controls. Pediatric Cardiology, 2020, 41, 1199-1205.	1.3	0
80	Phenotypic and molecular description of an individual with a disruptive variant in the SULF2 gene. Clinical Dysmorphology, 2020, 29, 144-147.	0.3	0
81	Case Report: SATB2-Associated Syndrome Overlapping With Clinical Mitochondrial Disease Presentation: Report of Two Cases. Frontiers in Genetics, 2021, 12, 692087.	2.3	0
82	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
83	Introduction to the special issue on Clinical Genetics in Latin America. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 873-875.	1.6	0