Yuri A Zarate

List of Publications by Year
in descending order

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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	Craniosynostosis is a feature of Costello syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 1280-1286.	1.2	3
2	A clinical scoring system for early onset (neonatal) Marfan syndrome. Genetics in Medicine, 2022, , .	2.4	1
3	Prevalence and Outcomes of Primary Left Ventricular Dysfunction in Marfan Syndrome. American Journal of Cardiology, 2022, 175, 119-126.	1.6	1
4	The landscape of submicroscopic structural variants at the OPN1LW/OPN1MW gene cluster on Xq28 underlying blue cone monochromacy. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	2
5	Growth in individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2952-2957.	1.2	3
6	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
7	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
8	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
9	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
10	Growth, development, and phenotypic spectrum of individuals with deletions of 2q33.1 involving SATB2. Clinical Genetics, 2021, 99, 547-557.	2.0	13
11	Individuals with SATB2-associated syndrome with and without autism have a recognizable metabolic profile and distinctive cellular energy metabolism alterations. Metabolic Brain Disease, 2021, 36, 1049-1056.	2.9	4
12	SATB2 â€associated syndrome in adolescents and adults. American Journal of Medical Genetics, Part A, 2021, 185, 2391-2398.	1.2	4
13	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
14	Case Report: SATB2-Associated Syndrome Overlapping With Clinical Mitochondrial Disease Presentation: Report of Two Cases. Frontiers in Genetics, 2021, 12, 692087.	2.3	0
15	Genetic and phenotypic heterogeneity in KIAA0753 â€related ciliopathies. American Journal of Medical Genetics, Part A, 2021, , .	1.2	2
16	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
17	CDK19-related disorder results from both loss-of-function and gain-of-function de novo missense variants. Genetics in Medicine, 2021, 23, 1050-1057.	2.4	7
18	Behavioral phenotype and sleep problems in SATB 2 â€associated syndrome. Developmental Medicine and Child Neurology, 2020, 62, 827-832.	2.1	8

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19	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
20	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. European Journal of Medical Genetics, 2020, 63, 103817.	1.3	6
21	Aortic Geometry in Patients with Duplication 7q11.23 Compared to Healthy Controls. Pediatric Cardiology, 2020, 41, 1199-1205.	1.3	0
22	Managing Sleep and Behavioral Problems in a Preschooler with SATB2-Associated Syndrome. Case Reports in Genetics, 2020, 2020, 1-4.	0.2	1
23	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
24	Epilepsy and Electroencephalographic Abnormalities in SATB2-Associated Syndrome. Pediatric Neurology, 2020, 112, 94-100.	2.1	10
25	Activating variants in <scp>PDGFRB</scp> result in a spectrum of disorders responsive to imatinib monotherapy. American Journal of Medical Genetics, Part A, 2020, 182, 1576-1591.	1.2	21
26	Phenotypic and molecular description of an individual with a disruptive variant in the SULF2 gene. Clinical Dysmorphology, 2020, 29, 144-147.	0.3	0
27	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
28	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
29	Speech, language, and feeding phenotypes of SATB2â€associated syndrome. Clinical Genetics, 2019, 96, 485-492.	2.0	10
30	Satb2 regulates proliferation and nuclear integrity of pre-osteoblasts. Bone, 2019, 127, 488-498.	2.9	14
31	Nuclear radiation and prevalence of structural birth defects among infants born to women from the Marshall Islands. Birth Defects Research, 2019, 111, 1192-1204.	1.5	11
32	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	61
33	Using facial analysis technology in a typical genetic clinic: experience from 30 individuals from a single institution. Journal of Human Genetics, 2019, 64, 1243-1245.	2.3	16
34	SATB2â€associated syndrome (SAS) and associated dental findings. Special Care in Dentistry, 2019, 39, 220-224.	0.8	10
35	Widening phenotypic spectrum of GABBR2 mutation. Acta Neurologica Belgica, 2019, 119, 493-496.	1.1	8
36	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38

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37	Frequency and age at occurrence of clinical manifestations of disease in patients with hypophosphatasia: a systematic literature review. Orphanet Journal of Rare Diseases, 2019, 14, 85.	2.7	21
38	Constitutive activation of the PI3Kâ€AKT pathway and cardiovascular abnormalities in an individual with Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1047-1052.	1.2	18
39	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
40	Less common underlying genetic diagnoses found in a cohort of 139 individuals surgically corrected for craniosynostosis. American Journal of Medical Genetics, Part A, 2018, 176, 487-491.	1.2	3
41	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	1.2	57
42	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
43	Ophthalmic findings in patients with arterial tortuosity syndrome and carriers: A case series. Ophthalmic Genetics, 2018, 39, 29-34.	1.2	9
44	Bone health and <scp>SATB2</scp>â€associated syndrome. Clinical Genetics, 2018, 93, 588-594.	2.0	18
45	Dental radiographic findings in 18 individuals with SATB2-associated syndrome. Clinical Oral Investigations, 2018, 22, 2947-2951.	3.0	7
46	An infant with ash-leaf and cafÃ© au lait spots: a case of double phakomatosis. Acta Neurologica Belgica, 2017, 117, 323-324.	1.1	3
47	Additional de novo missense genetic variants in <scp>NALCN</scp> associated with <scp>CLIFAHDD</scp> syndrome. Clinical Genetics, 2017, 91, 929-931.	2.0	10
48	Genotype and phenotype in 12 additional individuals with <scp>SATB2</scp>â€associated syndrome. Clinical Genetics, 2017, 92, 423-429.	2.0	33
49	Severe Metabolic Acidosis and Hepatopathy due to Leukoencephalopathy with Thalamus and Brainstem Involvement and High Lactate. Neuropediatrics, 2017, 48, 108-110.	0.6	4
50	Advanced cardiovascular imaging in Williams syndrome: Abnormalities, usefulness, and strategy for use. , 2017, 173, 1194-1199.		7
51	The role of IQSEC2 in syndromic intellectual disability: Narrowing the diagnostic odyssey. , 2017, 173, 2814-2820.		14
52	De novo variants in EBF3 are associated with hypotonia, developmental delay, intellectual disability, and autism. Journal of Physical Education and Sports Management, 2017, 3, a002097.	1.2	28
53	SATB2â€associated syndrome: Mechanisms, phenotype, and practical recommendations. American Journal of Medical Genetics, Part A, 2017, 173, 327-337.	1.2	81
54	SMARCE1, a rare cause of Coffinâ€“Siris Syndrome: Clinical description of three additional cases. American Journal of Medical Genetics, Part A, 2016, 170, 1967-1973.	1.2	18

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55	First clinical report of an infant with microcephaly and CASC5 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 2215-2218.	1.2	10
56	COL1A1 and COL1A2 sequencing results in cohort of patients undergoing evaluation for potential child abuse. American Journal of Medical Genetics, Part A, 2016, 170, 1858-1862.	1.2	19
57	Lessons from a pair of siblings with BPAN. European Journal of Human Genetics, 2016, 24, 1080-1083.	2.8	26
58	Aortic dilation, genetic testing, and associated diagnoses. Genetics in Medicine, 2016, 18, 356-363.	2.4	4
59	Further supporting evidence for the SATB2â€associated syndrome found through whole exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1026-1032.	1.2	42
60	Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. American Journal of Medical Genetics, Part A, 2015, 167, 2168-2175.	1.2	3
61	STAR syndrome is part of the differential diagnosis of females with anorectal malformations. American Journal of Medical Genetics, Part A, 2015, 167, 1940-1943.	1.2	13
62	Aortic dilation in pediatric patients. European Journal of Pediatrics, 2015, 174, 1585-1592.	2.7	9
63	Persistent congenital hyperinsulinism in two patients with Beckwith-Wiedemann syndrome due to mosaic uniparental disomy 11p. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 951-5.	0.9	5
64	Papillary thyroid cancer in a patient with interstitial 6q25 deletion including ARID1B. American Journal of Medical Genetics, Part A, 2014, 164, 1857-1859.	1.2	17
65	Unique Cerebrovascular Anomalies in Noonan Syndrome With RAF1 Mutation. Journal of Child Neurology, 2014, 29, NP13-NP17.	1.4	19
66	Severe neonatal presentation of Kleefstra syndrome in a patient with hypoplastic left heart syndrome and 9q34.3 microdeletion. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 985-990.	1.6	10
67	Description of the First Case of Adenomyomatosis of the Gallbladder in an Infant. Case Reports in Pediatrics, 2014, 2014, 1-3.	0.4	16
68	Cardiovascular and genitourinary anomalies in patients with duplications within the Williams syndrome critical region: Phenotypic expansion and review of the literature. American Journal of Medical Genetics, Part A, 2014, 164, 1998-2002.	1.2	22
69	Clinical utility of the Xâ€chromosome array. American Journal of Medical Genetics, Part A, 2013, 161, 120-130.	1.2	0
70	Description of another case of 3q26.33-3q27.2 microdeletion supports a recognizable phenotype. European Journal of Medical Genetics, 2013, 56, 624-625.	1.3	4
71	Genetic Causes of Macroglossia: Diagnostic Approach. Pediatrics, 2012, 129, e431-e437.	2.1	38
72	Infrequent Manifestations of Kabuki Syndrome in a Patient with Novel <i>MLL2</i> Mutation. Molecular Syndromology, 2012, 3, 180-184.	0.8	22

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73	47, XY, +der(Y),t(X;Y)(p21.1;p11.2): A unique case of XY sex reversal. American Journal of Medical Genetics, Part A, 2011, 155, 386-391.	1.2	4
74	Lethal presentation of neurofibromatosis and Noonan syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1360-1366.	1.2	17
75	Intestinal Mai rotation in a Patient with Pfeiffer Syndrome Type 2. Cleft Palate-Craniofacial Journal, 2010, 47, 638-641.	0.9	3
76	A case of minimal change disease in a Fabry patient. Pediatric Nephrology, 2010, 25, 553-556.	1.7	10
77	Evaluation of Growth in Patients With Isolated Cleft Lip and/or Cleft Palate. Pediatrics, 2010, 125, e543-e549.	2.1	22
78	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
79	Twinâ€“twin transfusion resulting in fetal cell contamination in Beckwithâ€“Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1569-1570.	1.2	2
80	Experience with hemihyperplasia and Beckwithâ€“Wiedemann syndrome surveillance protocol. American Journal of Medical Genetics, Part A, 2009, 149A, 1691-1697.	1.2	47
81	Fabry's disease. Lancet, The, 2008, 372, 1427-1435.	13.7	628
82	A new case of de novo 11q duplication in a patient with normal development and intelligence and review of the literature. American Journal of Medical Genetics, Part A, 2007, 143A, 265-270.	1.2	20
83	Novel Fibrillin 1 Mutation in a Case of Neonatal Marfan Syndrome: The Increasing Importance of Early Recognition. Congenital Heart Disease, 2007, 2, 342-346.	0.2	17