

Giuseppe Zampino

List of Publications by Year in descending order

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Version: 2024-02-01

45
papers

4,318
citations

430874

18
h-index

276875

41
g-index

45
all docs

45
docs citations

45
times ranked

4862
citing authors

#	ARTICLE	IF	CITATIONS
1	RASopathies and sigmoid-shaped ventricular septum morphology: evidence of a previously unappreciated cardiac phenotype. <i>Pediatric Research</i> , 2023, 93, 752-754.	2.3	1
2	Hyperactive HRAS dysregulates energetic metabolism in fibroblasts from patients with Costello syndrome via enhanced production of reactive oxidizing species. <i>Human Molecular Genetics</i> , 2022, 31, 561-575.	2.9	6
3	Genotypeâ€cardiac phenotype correlations in a large singleâ€center cohort of patients affected by RASopathies: Clinical implications and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 431-445.	1.2	25
4	Characterization of bone homeostasis in individuals affected by cardioâ€facioâ€cutaneous syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 414-421.	1.2	7
5	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. <i>European Journal of Medical Genetics</i> , 2022, 65, 104439.	1.3	5
6	Smith Magenis syndrome: First case of congenital heart defect in a patient with <i>Rai1</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2184-2186.	1.2	2
7	Bone tissue homeostasis and risk of fractures in Costello syndrome: A 4â€year followâ€up study. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 422-430.	1.2	5
8	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. <i>Obesity Surgery</i> , 2021, 31, 445-450.	2.1	2
9	Musculo-skeletal phenotype of Costello syndrome and cardio-facio-cutaneous syndrome: insights on the functional assessment status. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 43.	2.7	20
10	Smithâ€Magenis syndrome: Report of morphological and new functional cardiac findings with review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2003-2011.	1.2	5
11	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3153-3160.	1.2	3
12	Recognition Memory in Noonan Syndrome. <i>Brain Sciences</i> , 2021, 11, 169.	2.3	2
13	Crisponi/coldâ€induced sweating syndrome: Differential diagnosis, pathogenesis and treatment concepts. <i>Clinical Genetics</i> , 2020, 97, 209-221.	2.0	12
14	Treatment of Dystonia Using Trihexyphenidyl in Costello Syndrome. <i>Brain Sciences</i> , 2020, 10, 450.	2.3	4
15	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. <i>American Journal of Human Genetics</i> , 2020, 107, 499-513.	6.2	48
16	Impact of Costello syndrome on growth patterns. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2797-2799.	1.2	10
17	The dark side of <sc>COVID</sc>â€19: The need of integrated medicine for children with special care needs. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1988-1989.	1.2	8
18	One case of anetoderma postâ€vitamin K 1 injection in a newborn. <i>International Journal of Dermatology</i> , 2020, 59, e168-e169.	1.0	2

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19	Nissen fundoplication in Cornelia de Lange syndrome spectrum: Who are the potential candidates?. American Journal of Medical Genetics, Part A, 2020, 182, 1697-1703.	1.2	1
20	Phenotype evolution and health issues of adults with Beckwith-Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1691-1702.	1.2	21
21	Pain in individuals with RASopathies: Prevalence and clinical characterization in a sample of 80 affected patients. American Journal of Medical Genetics, Part A, 2019, 179, 940-947.	1.2	21
22	Cover Image, Volume 179A, Number 9, September 2019. , 2019, 179, i-i.		2
23	Psychopathological features in Noonan syndrome. European Journal of Paediatric Neurology, 2018, 22, 170-177.	1.6	26
24	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	6.2	73
25	Ausgedehnte, unregelmäßige Mongolenflecken als Hinweis auf GM1-Gangliosidose Typ 1. JDDG - Journal of the German Society of Dermatology, 2016, 14, 301-302.	0.8	0
26	Respiratory and gastrointestinal dysfunctions associated with auriculocondylar syndrome and a homozygous PLCB4 loss-of-function mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1471-1478.	1.2	12
27	Extensive irregular Mongolian blue spots as a clue for GM1 gangliosidosis type 1. JDDG - Journal of the German Society of Dermatology, 2016, 14, 301-302.	0.8	3
28	Recommendations of the Scientific Committee of the Italian Beckwith-Wiedemann Syndrome Association on the diagnosis, management and follow-up of the syndrome. European Journal of Medical Genetics, 2016, 59, 52-64.	1.3	76
29	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. Journal of Pediatrics, 2016, 170, 322-324.	1.8	35
30	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	2.5	67
31	Costello syndrome: Analysis of the posterior cranial fossa in children with posterior fossa crowding. Neuroradiology Journal, 2015, 28, 254-258.	1.2	12
32	Editorial: Care of adults with Down syndrome: Gaps and needs. European Journal of Internal Medicine, 2015, 26, 375-376.	2.2	16
33	Decreased bone mineral density in Costello syndrome. Molecular Genetics and Metabolism, 2014, 111, 41-45.	1.1	22
34	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. Journal of Medical Genetics, 2013, 50, 493-499.	3.2	40
35	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	2.5	97
36	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. American Journal of Human Genetics, 2010, 87, 250-257.	6.2	221

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37	Craniosynostosis in patients with Noonan syndrome caused by germline <i>KRAS</i> mutations. American Journal of Medical Genetics, Part A, 2009, 149A, 1036-1040.	1.2	46
38	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. European Journal of Human Genetics, 2009, 17, 733-740.	2.8	74
39	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	21.4	358
40	Diversity, parental germline origin, and phenotypic spectrum of de novo <i>HRAS</i> missense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	2.5	123
41	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	21.4	523
42	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	21.4	624
43	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	21.4	1,555
44	Costello syndrome: Further clinical delineation, natural history, genetic definition, and nosology. American Journal of Medical Genetics Part A, 1993, 47, 176-183.	2.4	96
45	Multidisciplinary Management of Costello Syndrome: Current Perspectives. Journal of Multidisciplinary Healthcare, 0, Volume 15, 1277-1296.	2.7	7