Giuseppe Zampino

List of Publications by Citations

42 papers	3,466 citations	16 h-index	45 g-index
45 ext. papers	4,009 ext. citations	6.5 avg, IF	3.81 L-index

#	Paper	IF	Citations
42	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. *Nature Genetics*, **2001**, 29, 465-8	36.3	1312
41	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. *Nature Genetics*, **2007**, 39, 1007-12	36.3	523
40	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. *Nature Genetics*, **2007**, 39, 75-9	36.3	440
39	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. *Nature Genetics*, **2009**, 41, 1022-6	36.3	291
38	Heterozygous germline mutations in the CBL tumor-suppressor gene cause a Noonan syndrome-like phenotype. *American Journal of Human Genetics*, **2010**, 87, 250-7	11	179
37	Diversity, parental germline origin, and phenotypic spectrum of de novo HRAS missense changes in Costello syndrome. *Human Mutation*, **2007**, 28, 265-72	4.7	104
36	Costello syndrome: further clinical delineation, natural history, genetic definition, and nosology. *American Journal of Medical Genetics Part A*, **1993**, 47, 176-83		86
35	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. *Human Mutation*, **2011**, 32, 760-72	4.7	82
34	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. *European Journal of Human Genetics*, **2009**, 17, 733-40	5.3	57
33	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	2.6	56
32	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. *Human Mutation*, **2015**, 36, 1080-7	4.7	51
31	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2018**, 103, 621-630	11	45
30	Craniosynostosis in patients with Noonan syndrome caused by germline KRAS mutations. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1036-40	2.5	40
29	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. *Journal of Medical Genetics*, **2013**, 50, 493-9	5.8	33
28	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
27	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. *Journal of Pediatrics*, **2016**, 170, 322-4	3.6	24
26	Decreased bone mineral density in Costello syndrome. *Molecular Genetics and Metabolism*, **2014**, 111, 41-5	3.7	15

25	Psychopathological features in Noonan syndrome. *European Journal of Paediatric Neurology*, **2018**, 22, 170-177	3.8	13
24	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	2.5	11
23	Costello syndrome: Analysis of the posterior cranial fossa in children with posterior fossa crowding. *Neuroradiology Journal*, **2015**, 28, 254-8	2	8
22	Musculo-skeletal phenotype of Costello syndrome and cardio-facio-cutaneous syndrome: insights on the functional assessment status. *Orphanet Journal of Rare Diseases*, **2021**, 16, 43	4.2	7
21	The dark side of COVID-19: The need of integrated medicine for children with special care needs. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1988-1989	2.5	6
20	Phenotype evolution and health issues of adults with Beckwith-Wiedemann syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1691-1702	2.5	6
19	Crisponi/cold-induced sweating syndrome: Differential diagnosis, pathogenesis and treatment concepts. *Clinical Genetics*, **2020**, 97, 209-221	4	6
18	Respiratory and gastrointestinal dysfunctions associated with auriculo-condylar syndrome and a homozygous PLCB4 loss-of-function mutation. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1471-8	2.5	6
17	Impact of Costello syndrome on growth patterns. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2797-2799	2.5	5
16	Treatment of Dystonia Using Trihexyphenidyl in Costello Syndrome. *Brain Sciences*, **2020**, 10,	3.4	3
15	Extensive irregular Mongolian blue spots as a clue for GM1 gangliosidosis type 1. *JDDG - Journal of the German Society of Dermatology*, **2016**, 14, 301-2	1.2	3
14	One case of anetoderma post-vitamin K injection in a newborn. *International Journal of Dermatology*, **2020**, 59, e168-e169	1.7	2
13	Genotype-cardiac phenotype correlations in a large single-center cohort of patients affected by RASopathies: Clinical implications and literature review. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	2
12	Smith-Magenis syndrome: Report of morphological and new functional cardiac findings with review of the literature. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2003-2011	2.5	2
11	Cover Image, Volume 179A, Number 9, September 2019 **2019**, 179, i-i		2
10	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. *Obesity Surgery*, **2021**, 31, 445-450	3.7	2
9	Hyperactive HRAS dysregulates energetic metabolism in fibroblasts from patients with Costello syndrome via enhanced production of reactive oxidizing species. *Human Molecular Genetics*, **2021**,	5.6	2
8	Characterization of bone homeostasis in individuals affected by cardio-facio-cutaneous syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 188, 414	2.5	1

7	Nissen fundoplication in Cornelia de Lange syndrome spectrum: Who are the potential candidates?. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1697-1703	2.5	1
6	Bone tissue homeostasis and risk of fractures in Costello syndrome: A 4-year follow-up study.. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	1
5	Metabolic profiling of Costello syndrome: Insights from a single-center cohort.. *European Journal of Medical Genetics*, **2022**, 104439	2.6	0
4	Broadening the phenotypic spectrum of Beta3GalT6-associated phenotypes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3153-3160	2.5	0
3	Smith Magenis syndrome: First case of congenital heart defect in a patient with Rai1 mutation.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
2	Multidisciplinary Management of Costello Syndrome: Current Perspectives. *Journal of Multidisciplinary Healthcare*,Volume 15, 1277-1296	2.8	0
1	Ausgedehnte, unregelmße Mongolenflecken als Hinweis auf GM1-Gangliosidose Typ 1. *JDDG - Journal of the German Society of Dermatology*, **2016**, 14, 301-302	1.2	