Giuseppe Zampino

List of Publications by Year
in descending order

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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	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	21.4	1,555
2	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	21.4	624
3	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	21.4	523
4	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
5	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
6	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	2.5	123
7	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
8	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
9	Recommendations of the Scientific Committee of the Italian Beckwithâ€“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
10	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2009, 17, 733-740.	2.8	74
11	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	6.2	73
12	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	2.5	67
13	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
14	Craniosynostosis in patients with Noonan syndrome caused by germline KRAS mutations. American Journal of Medical Genetics, Part A, 2009, 149A, 1036-1040.	1.2	46
15	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
16	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. Journal of Pediatrics, 2016, 170, 322-324.	1.8	35
17	Psychopathological features in Noonan syndrome. European Journal of Paediatric Neurology, 2018, 22, 170-177.	1.6	26
18	Genotypeâ€cardiac phenotype correlations in a large singleâ€center cohort of patients affected by RASopathies: Clinical implications and literature review. American Journal of Medical Genetics, Part A, 2022, 188, 431-445.	1.2	25

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19	Decreased bone mineral density in Costello syndrome. Molecular Genetics and Metabolism, 2014, 111, 41-45.	1.1	22
20	Phenotype evolution and health issues of adults with Beckwithâ€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	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.	2.7	20
23	Editorial: Care of adults with Down syndrome: Gaps and needs. European Journal of Internal Medicine, 2015, 26, 375-376.	2.2	16
24	Costello syndrome: Analysis of the posterior cranial fossa in children with posterior fossa crowding. Neuroradiology Journal, 2015, 28, 254-258.	1.2	12
25	Respiratory and gastrointestinal dysfunctions associated with auriculoâ€condylar syndrome and a homozygous PLCB4 lossâ€ofâ€function mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1471-1478.	1.2	12
26	Crisponi/coldâ€induced sweating syndrome: Differential diagnosis, pathogenesis and treatment concepts. Clinical Genetics, 2020, 97, 209-221.	2.0	12
27	Impact of Costello syndrome on growth patterns. American Journal of Medical Genetics, Part A, 2020, 182, 2797-2799.	1.2	10
28	The dark side of <scp>COVID</scp>â€19: The need of integrated medicine for children with special care needs. American Journal of Medical Genetics, Part A, 2020, 182, 1988-1989.	1.2	8
29	Characterization of bone homeostasis in individuals affected by cardioâ€facioâ€cutaneous syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 414-421.	1.2	7
30	Multidisciplinary Management of Costello Syndrome: Current Perspectives. Journal of Multidisciplinary Healthcare, 0, Volume 15, 1277-1296.	2.7	7
31	Hyperactive HRAS dysregulates energetic metabolism in fibroblasts from patients with Costello syndrome via enhanced production of reactive oxidizing species. Human Molecular Genetics, 2022, 31, 561-575.	2.9	6
32	Smithâ€“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.	1.2	5
33	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. European Journal of Medical Genetics, 2022, 65, 104439.	1.3	5
34	Bone tissue homeostasis and risk of fractures in Costello syndrome: A 4â€year followâ€up study. American Journal of Medical Genetics, Part A, 2022, 188, 422-430.	1.2	5
35	Treatment of Dystonia Using Trihexyphenidyl in Costello Syndrome. Brain Sciences, 2020, 10, 450.	2.3	4
36	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

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37	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3153-3160.	1.2	3
38	Cover Image, Volume 179A, Number 9, September 2019. , 2019, 179, i-i.		2
39	One case of anetoderma postâ€vitamin K 1 injection in a newborn. International Journal of Dermatology, 2020, 59, e168-e169.	1.0	2
40	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. Obesity Surgery, 2021, 31, 445-450.	2.1	2
41	Recognition Memory in Noonan Syndrome. Brain Sciences, 2021, 11, 169.	2.3	2
42	Smith Magenis syndrome: First case of congenital heart defect in a patient with Rai1 mutation. American Journal of Medical Genetics, Part A, 2022, 188, 2184-2186.	1.2	2
43	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
44	RASopathies and sigmoid-shaped ventricular septum morphology: evidence of a previously unappreciated cardiac phenotype. Pediatric Research, 2023, 93, 752-754.	2.3	1
45	Ausgedehnte, unregelmÃÃŸige Mongolenflecken als Hinweis auf GM1-Gangliosidose Typ 1. JDDG - Journal of the German Society of Dermatology, 2016, 14, 301-302.	0.8	0