Roberta Onesimo

List of Publications by Year
in descending order

66	918	566801 15	552369 26
papers	citations	h-index	g-index

67	67	67	1323
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A multicentre retrospective study of 66 <scp>I</scp>talian children with food proteinâ€induced enterocolitis syndrome: different management for different phenotypes. Clinical and Experimental Allergy, 2012, 42, 1257-1265.	1.4	179
2	Responsiveness to intravenous immunoglobulins and occurrence of coronary artery abnormalities in a single-center cohort of Italian patients with Kawasaki syndrome. Rheumatology International, 2010, 30, 841-846.	1.5	57
3	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. American Journal of Human Genetics, 2020, 107, 1129-1148.	2.6	38
4	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. Journal of Pediatrics, 2016, 170, 322-324.	0.9	35
5	Chromosome 9p deletion syndrome and sex reversal: Novel findings and redefinition of the critically deleted regions. American Journal of Medical Genetics, Part A, 2012, 158A, 2266-2271.	0.7	33
6	First evidence of a therapeutic effect of miransertib in a teenager with Proteus syndrome and ovarian carcinoma. American Journal of Medical Genetics, Part A, 2019, 179, 1319-1324.	0.7	33
7	Congenital immunodeficiency in an individual with Wiedemannâ€“Steiner syndrome due to a novel missense mutation in KMT2A. American Journal of Medical Genetics, Part A, 2016, 170, 2389-2393.	0.7	29
8	Brain CT scan for pediatric minor accidental head injury. An Italian experience and review of literature. Child's Nervous System, 2012, 28, 1063-1068.	0.6	25
9	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.	0.7	25
10	Incomplete Kawasaki syndrome followed by systemic onset-juvenile idiopathic arthritis mimicking Kawasaki syndrome. Rheumatology International, 2010, 30, 535-539.	1.5	23
11	Specific oral tolerance induction (SOTI) in pediatric age: Clinical research or just routine practice?. Pediatric Allergy and Immunology, 2010, 21, e446-9.	1.1	22
12	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.	0.7	21
13	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	1.0	21
14	Isolated contact urticaria caused by immunoglobulin E-mediated fish allergy. Israel Medical Association Journal, 2012, 14, 11-3.	0.1	21
15	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.	1.2	20
16	A quality evaluation methodology of health web-pages for non-professionals. Informatics for Health and Social Care, 2004, 29, 95-107.	1.0	19
17	Ultrasound assessment of diaphragmatic function in type 1 spinal muscular atrophy. Pediatric Pulmonology, 2020, 55, 1781-1788.	1.0	18
18	Oral and Swallowing Abilities Tool (OrSAT) for Type 1 SMA Patients: Development of a New Module. Journal of Neuromuscular Diseases, 2021, 8, 589-601.	1.1	16

#	Article	IF	Citations
19	Genotypes and phenotypes heterogeneity in PIK3CA-related overgrowth spectrum and overlapping conditions: 150 novel patients and systematic review of 1007 patients with PIK3CA pathogenetic variants. Journal of Medical Genetics, 2023, 60, 163-173.	1.5	15
20	Measles-induced respiratory distress, air-leak and ARDS. European Journal of Clinical Microbiology and Infectious Diseases, 2010, 29, 181-185.	1.3	14
21	Childrenâ€™s Healthcare During Corona Virus Disease 19 Pandemic. Pediatric Infectious Disease Journal, 2020, 39, e137-e140.	1.1	14
22	Contactless: a new personalised telehealth model in chronic pediatric diseases and disability during the COVID-19 era. Italian Journal of Pediatrics, 2021, 47, 29.	1.0	13
23	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. Genes, 2021, 12, 1316.	1.0	13
24	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.	0.7	12
25	Visual Function and Ophthalmological Findings in CHARGE Syndrome: Revision of Literature, Definition of a New Clinical Spectrum and Genotype Phenotype Correlation. Genes, 2021, 12, 972.	1.0	12
26	Food-dependent exercise-induced anaphylaxis (FDEIA) by nectarine in a paediatric patient with weakly positive nectarine prick-by-prick and negative specific IgE to Pru p 3. Allergologia Et Immunopathologia, 2013, 41, 201-203.	1.0	10
27	The re-emergence of dengue virus in non-endemic countries: a case series. BMC Research Notes, 2014, 7, 596.	0.6	10
28	Impact of Costello syndrome on growth patterns. American Journal of Medical Genetics, Part A, 2020, 182, 2797-2799.	0.7	10
29	Biallelic TRNT1 variants in a child with B cell immunodeficiency, periodic fever and developmental delay without sideroblastic anemia (SIFD variant). Immunology Letters, 2020, 225, 64-65.	1.1	10
30	IVIG treatment for VZV-related acute inflammatory polyneuropathy in a child. BMJ Case Reports, 2012, 2012, bcr2012006362-bcr2012006362.	0.2	8
31	Prevalence of adverse reactions following a passed oral food challenge and factors affecting successful re-introduction of foods. A retrospective study of a cohort of 199 children. Allergologia Et Immunopathologia, 2016, 44, 54-58.	1.0	8
32	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.	0.7	8
33	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	0.7	7
34	Is heel prick as safe as we think?. BMJ Case Reports, 2011, 2011, bcr0820114677-bcr0820114677.	0.2	7
35	Characterization of bone homeostasis in individuals affected by cardioâ€facioâ€cutaneous syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 414-421.	0.7	7
36	Montelukast versus inhaled corticosteroids as monotherapy for prevention of asthma: which one is best?. Allergologia Et Immunopathologia, 2009, 37, 26-30.	1.0	6

#	Article	IF	Citations
37	Old treatments for new genetic conditions: Sirolimus therapy in a child affected by mosaic overgrowth with fibroadipose hyperplasia. Clinical Genetics, 2019, 96, 102-103.	1.0	6
38	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.	1.4	6
39	Hypoglycaemia in patients with type 1 SMA: an underdiagnosed problem?. Archives of Disease in Childhood, 2020, 105, 707-707.	1.0	6
40	Intravenous Immunoglobulin therapy for anti-E hemolytic disease in the newborn. Journal of Maternal-Fetal and Neonatal Medicine, 2010, 23, 1059-1061.	0.7	5
41	Pott's puffy tumour by Streptoccocus intermedius a rare complication of sinusitis. BMJ Case Reports, 2011, 2011, bcr0820114660-bcr0820114660.	0.2	5
42	Oligonephronia and Wolfâ€Hirschhorn syndrome: A further observation. American Journal of Medical Genetics, Part A, 2018, 176, 409-414.	0.7	5
43	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.	0.7	5
44	The Evolution of Web-based Medical Information on Sore Throat: a Longitudinal Study. Journal of Medical Internet Research, 2003, 5, e10.	2.1	5
45	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. European Journal of Medical Genetics, 2022, 65, 104439.	0.7	5
46	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.	0.7	5
47	Treatment of Dystonia Using Trihexyphenidyl in Costello Syndrome. Brain Sciences, 2020, 10, 450.	1.1	4
48	Immunoglobulin deficiency associated with a MAP2K1-related mutation causing cardio-facio-cutaneous syndrome. Immunology Letters, 2020, 227, 79-80.	1.1	4
49	The tuberculosis spectrum: Translating basic research into pediatric clinical practice. Medical Hypotheses, 2020, 141, 108091.	0.8	4
50	Intestinal Permeability in Children with Functional Gastrointestinal Disorders: The Effects of Diet. Nutrients, 2022, 14, 1578.	1.7	4
51	Short Therapy in a Septic Arthritis of the Neonatal Hip. Mental Illness, 2019, 11, 8161.	0.8	3
52	Two case reports of fetal alcohol syndrome: broadening into the spectrum of cardiac disease to personalize and to improve clinical assessment. Italian Journal of Pediatrics, 2019, 45, 167.	1.0	3
53	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3153-3160.	0.7	3
54	Prevalence of bladder cancer in Costello syndrome: New insights to drive clinical decisionâ€making. Clinical Genetics, 2022, 101, 454-458.	1.0	3

#	Article	IF	Citations
55	Oral and Swallowing Abilities Tool (OrSAT) in nusinersen treated patients. Archives of Disease in Childhood, 2022, 107, 912-916.	1.0	3
56	Risk of adverse IgE-mediate reaction at the first egg ingestion in children with atopic dermatitis. Results of a case-control study. Allergologia Et Immunopathologia, 2014, 42, 96-101.	1.0	2
57	One case of anetoderma postâ€vitamin K 1 injection in a newborn. International Journal of Dermatology, 2020, 59, e168-e169.	0.5	2
58	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. Obesity Surgery, 2021, 31, 445-450.	1.1	2
59	Enlarged spinal nerve roots in RASopathies: Report of two cases. European Journal of Medical Genetics, 2021, 64, 104187.	0.7	2
60	Congenital syphilis: remember to not forget. BMJ Case Reports, 2012, 2012, .	0.2	2
61	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.	0.7	2
62	The influence of quality criteria on parents' evaluation of medical web-pages: An Italian randomised trial. Technology and Health Care, 2007, 15, 399-406.	0.5	1
63	Basedow-Gravesâ€™ disease in a pediatric patient with Sticlker syndrome, a new endocrine finding to improve personalized treatment. Italian Journal of Pediatrics, 2020, 46, 178.	1.0	1
64	Nissen fundoplication in Cornelia de Lange syndrome spectrum: Who are the potential candidates?. American Journal of Medical Genetics, Part A, 2020, 182, 1697-1703.	0.7	1
65	A crying baby: not simply infant colic. BMJ Case Reports, 2012, 2012, bcr2012006544-bcr2012006544.	0.2	0
66	Don't forget 'simple' causes of abdominal pain. BMJ Case Reports, 2012, 2012, bcr2012006502-bcr2012006502.	0.2	0