Roberta Onesimo

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8678776/publications.pdf

Version: 2024-02-01

66 papers	918 citations	567281 15 h-index	552781 26 g-index
67 all docs	67 docs citations	67 times ranked	1323 citing authors

#	Article	IF	Citations
1	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.	3.2	15
2	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
3	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
4	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
5	Prevalence of bladder cancer in Costello syndrome: New insights to drive clinical decisionâ€making. Clinical Genetics, 2022, 101, 454-458.	2.0	3
6	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. European Journal of Medical Genetics, 2022, 65, 104439.	1.3	5
7	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
8	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
9	Intestinal Permeability in Children with Functional Gastrointestinal Disorders: The Effects of Diet. Nutrients, 2022, 14, 1578.	4.1	4
10	Oral and Swallowing Abilities Tool (OrSAT) in nusinersen treated patients. Archives of Disease in Childhood, 2022, 107, 912-916.	1.9	3
11	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. Obesity Surgery, 2021, 31, 445-450.	2.1	2
12	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
13	Contactless: a new personalised telehealth model in chronic pediatric diseases and disability during the COVID-19 era. Italian Journal of Pediatrics, 2021, 47, 29.	2.6	13
14	Enlarged spinal nerve roots in RASopathies: Report of two cases. European Journal of Medical Genetics, 2021, 64, 104187.	1.3	2
15	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
16	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
17	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.	2.4	12
18	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3153-3160.	1.2	3

#	Article	IF	Citations
19	Oral and Swallowing Abilities Tool (OrSAT) for Type 1 SMA Patients: Development of a New Module. Journal of Neuromuscular Diseases, 2021, 8, 589-601.	2.6	16
20	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. Genes, 2021, 12, 1316.	2.4	13
21	Childrenâ€™s Healthcare During Corona Virus Disease 19 Pandemic. Pediatric Infectious Disease Journal, 2020, 39, e137-e140.	2.0	14
22	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.	2.6	1
23	Treatment of Dystonia Using Trihexyphenidyl in Costello Syndrome. Brain Sciences, 2020, 10, 450.	2.3	4
24	Immunoglobulin deficiency associated with a MAP2K1-related mutation causing cardio-facio-cutaneous syndrome. Immunology Letters, 2020, 227, 79-80.	2.5	4
25	Impact of Costello syndrome on growth patterns. American Journal of Medical Genetics, Part A, 2020, 182, 2797-2799.	1.2	10
26	Ultrasound assessment of diaphragmatic function in type 1 spinal muscular atrophy. Pediatric Pulmonology, 2020, 55, 1781-1788.	2.0	18
27	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
28	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.	2.5	10
29	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	1.3	7
30	One case of anetoderma postâ€vitamin K 1 injection in a newborn. International Journal of Dermatology, 2020, 59, e168-e169.	1.0	2
31	The tuberculosis spectrum: Translating basic research into pediatric clinical practice. Medical Hypotheses, 2020, 141, 108091.	1.5	4
32	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. American Journal of Human Genetics, 2020, 107, 1129-1148.	6.2	38
33	Hypoglycaemia in patients with type 1 SMA: an underdiagnosed problem?. Archives of Disease in Childhood, 2020, 105, 707-707.	1.9	6
34	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
35	Short Therapy in a Septic Arthritis of the Neonatal Hip. Mental Illness, 2019, 11, 8161.	0.8	3
36	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.	1.2	33

#	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.	2.0	6
38	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
39	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.	2.6	3
40	Oligonephronia and Wolfâ€Hirschhorn syndrome: A further observation. American Journal of Medical Genetics, Part A, 2018, 176, 409-414.	1.2	5
41	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.	1.2	29
42	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
43	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. Journal of Pediatrics, 2016, 170, 322-324.	1.8	35
44	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.	1.7	8
45	The re-emergence of dengue virus in non-endemic countries: a case series. BMC Research Notes, 2014, 7, 596.	1.4	10
46	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.	1.7	2
47	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.	1.7	10
48	IVIG treatment for VZV-related acute inflammatory polyneuropathy in a child. BMJ Case Reports, 2012, 2012, bcr2012006362-bcr2012006362.	0.5	8
49	A crying baby: not simply infant colic. BMJ Case Reports, 2012, 2012, bcr2012006544-bcr2012006544.	0.5	0
50	Don't forget 'simple' causes of abdominal pain. BMJ Case Reports, 2012, 2012, bcr2012006502-bcr2012006502.	0.5	0
51	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.	1.2	33
52	Brain CT scan for pediatric minor accidental head injury. An Italian experience and review of literature. Child's Nervous System, 2012, 28, 1063-1068.	1.1	25
53	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.	2.9	179
54	Congenital syphilis: remember to not forget. BMJ Case Reports, 2012, 2012, .	0.5	2

#	Article	IF	Citations
55	Isolated contact urticaria caused by immunoglobulin E-mediated fish allergy. Israel Medical Association Journal, 2012, 14, 11-3.	0.1	21
56	Pott's puffy tumour by Streptoccocus intermedius a rare complication of sinusitis. BMJ Case Reports, 2011, 2011, bcr0820114660-bcr0820114660.	0.5	5
57	Is heel prick as safe as we think?. BMJ Case Reports, 2011, 2011, bcr0820114677-bcr0820114677.	0.5	7
58	Incomplete Kawasaki syndrome followed by systemic onset-juvenile idiopathic arthritis mimicking Kawasaki syndrome. Rheumatology International, 2010, 30, 535-539.	3.0	23
59	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.	3.0	57
60	Measles-induced respiratory distress, air-leak and ARDS. European Journal of Clinical Microbiology and Infectious Diseases, 2010, 29, 181-185.	2.9	14
61	Specific oral tolerance induction (SOTI) in pediatric age: Clinical research or just routine practice?. Pediatric Allergy and Immunology, 2010, 21, e446-9.	2.6	22
62	Intravenous Immunoglobulin therapy for anti-E hemolytic disease in the newborn. Journal of Maternal-Fetal and Neonatal Medicine, 2010, 23, 1059-1061.	1.5	5
63	Montelukast versus inhaled corticosteroids as monotherapy for prevention of asthma: which one is best?. Allergologia Et Immunopathologia, 2009, 37, 26-30.	1.7	6
64	The influence of quality criteria on parents' evaluation of medical web-pages: An Italian randomised trial. Technology and Health Care, 2007, 15, 399-406.	1.2	1
65	A quality evaluation methodology of health web-pages for non-professionals. Informatics for Health and Social Care, 2004, 29, 95-107.	1.0	19
66	The Evolution of Web-based Medical Information on Sore Throat: a Longitudinal Study. Journal of Medical Internet Research, 2003, 5, e10.	4.3	5