

# 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

566801

15  
h-index

552369

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. <i>Journal of Medical Genetics</i> , 2023, 60, 163-173.	1.5	15
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.	1.4	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.	0.7	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.	0.7	7
5	Prevalence of bladder cancer in Costello syndrome: New insights to drive clinical decisionâ€making. <i>Clinical Genetics</i> , 2022, 101, 454-458.	1.0	3
6	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. <i>European Journal of Medical Genetics</i> , 2022, 65, 104439.	0.7	5
7	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.	0.7	2
8	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.	0.7	5
9	Intestinal Permeability in Children with Functional Gastrointestinal Disorders: The Effects of Diet. <i>Nutrients</i> , 2022, 14, 1578.	1.7	4
10	Oral and Swallowing Abilities Tool (OrSAT) in nusinersen treated patients. <i>Archives of Disease in Childhood</i> , 2022, 107, 912-916.	1.0	3
11	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. <i>Obesity Surgery</i> , 2021, 31, 445-450.	1.1	2
12	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.	1.2	20
13	Contactless: a new personalised telehealth model in chronic pediatric diseases and disability during the COVID-19 era. <i>Italian Journal of Pediatrics</i> , 2021, 47, 29.	1.0	13
14	Enlarged spinal nerve roots in RASopathies: Report of two cases. <i>European Journal of Medical Genetics</i> , 2021, 64, 104187.	0.7	2
15	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.	0.7	5
16	<i>ANKRD11</i> variants: <i>KBG</i> syndrome and beyond. <i>Clinical Genetics</i> , 2021, 100, 187-200.	1.0	21
17	Visual Function and Ophthalmological Findings in CHARGE Syndrome: Revision of Literature, Definition of a New Clinical Spectrum and Genotype Phenotype Correlation. <i>Genes</i> , 2021, 12, 972.	1.0	12
18	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3153-3160.	0.7	3

#	ARTICLE	IF	CITATIONS
19	Oral and Swallowing Abilities Tool (OrSAT) for Type 1 SMA Patients: Development of a New Module. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 589-601.	1.1	16
20	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. <i>Genes</i> , 2021, 12, 1316.	1.0	13
21	Children's Healthcare During Corona Virus Disease 19 Pandemic. <i>Pediatric Infectious Disease Journal</i> , 2020, 39, e137-e140.	1.1	14
22	Basedow-Graves disease in a pediatric patient with Sticler syndrome, a new endocrine finding to improve personalized treatment. <i>Italian Journal of Pediatrics</i> , 2020, 46, 178.	1.0	1
23	Treatment of Dystonia Using Trihexyphenidyl in Costello Syndrome. <i>Brain Sciences</i> , 2020, 10, 450.	1.1	4
24	Immunoglobulin deficiency associated with a MAP2K1-related mutation causing cardio-facio-cutaneous syndrome. <i>Immunology Letters</i> , 2020, 227, 79-80.	1.1	4
25	Impact of Costello syndrome on growth patterns. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2797-2799.	0.7	10
26	Ultrasound assessment of diaphragmatic function in type 1 spinal muscular atrophy. <i>Pediatric Pulmonology</i> , 2020, 55, 1781-1788.	1.0	18
27	The dark side of COVID-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.	0.7	8
28	Biallelic TRNT1 variants in a child with B cell immunodeficiency, periodic fever and developmental delay without sideroblastic anemia (SIFD variant). <i>Immunology Letters</i> , 2020, 225, 64-65.	1.1	10
29	CantÃ© syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. <i>European Journal of Medical Genetics</i> , 2020, 63, 103996.	0.7	7
30	One case of anetoderma postvitamin K 1 injection in a newborn. <i>International Journal of Dermatology</i> , 2020, 59, e168-e169.	0.5	2
31	The tuberculosis spectrum: Translating basic research into pediatric clinical practice. <i>Medical Hypotheses</i> , 2020, 141, 108091.	0.8	4
32	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. <i>American Journal of Human Genetics</i> , 2020, 107, 1129-1148.	2.6	38
33	Hypoglycaemia in patients with type 1 SMA: an underdiagnosed problem?. <i>Archives of Disease in Childhood</i> , 2020, 105, 707-707.	1.0	6
34	Nissen fundoplication in Cornelia de Lange syndrome spectrum: Who are the potential candidates?. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1697-1703.	0.7	1
35	Short Therapy in a Septic Arthritis of the Neonatal Hip. <i>Mental Illness</i> , 2019, 11, 8161.	0.8	3
36	First evidence of a therapeutic effect of miransertib in a teenager with Proteus syndrome and ovarian carcinoma. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1319-1324.	0.7	33

#	ARTICLE	IF	CITATIONS
37	Old treatments for new genetic conditions: Sirolimus therapy in a child affected by mosaic overgrowth with fibroadipose hyperplasia. <i>Clinical Genetics</i> , 2019, 96, 102-103.	1.0	6
38	Pain in individuals with RASopathies: Prevalence and clinical characterization in a sample of 80 affected patients. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 940-947.	0.7	21
39	Two case reports of fetal alcohol syndrome: broadening into the spectrum of cardiac disease to personalize and to improve clinical assessment. <i>Italian Journal of Pediatrics</i> , 2019, 45, 167.	1.0	3
40	Oligonephronia and Wolfâ€“Hirschhorn syndrome: A further observation. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 409-414.	0.7	5
41	Congenital immunodeficiency in an individual with Wiedemannâ€“Steiner syndrome due to a novel missense mutation in <i>KMT2A</i> . <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2389-2393.	0.7	29
42	Respiratory and gastrointestinal dysfunctions associated with auriculoâ€“condylar syndrome and a homozygous <i>PLCB4</i> lossâ€“ofâ€“function mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1471-1478.	0.7	12
43	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. <i>Journal of Pediatrics</i> , 2016, 170, 322-324.	0.9	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. <i>Allergologia Et Immunopathologia</i> , 2016, 44, 54-58.	1.0	8
45	The re-emergence of dengue virus in non-endemic countries: a case series. <i>BMC Research Notes</i> , 2014, 7, 596.	0.6	10
46	Risk of adverse IgE-mediate reaction at the first egg ingestion in children with atopic dermatitis. Results of a case-control study. <i>Allergologia Et Immunopathologia</i> , 2014, 42, 96-101.	1.0	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. <i>Allergologia Et Immunopathologia</i> , 2013, 41, 201-203.	1.0	10
48	IVIg treatment for VZV-related acute inflammatory polyneuropathy in a child. <i>BMJ Case Reports</i> , 2012, 2012, bcr2012006362-bcr2012006362.	0.2	8
49	A crying baby: not simply infant colic. <i>BMJ Case Reports</i> , 2012, 2012, bcr2012006544-bcr2012006544.	0.2	0
50	Don't forget 'simple' causes of abdominal pain. <i>BMJ Case Reports</i> , 2012, 2012, bcr2012006502-bcr2012006502.	0.2	0
51	Chromosome 9p deletion syndrome and sex reversal: Novel findings and redefinition of the critically deleted regions. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2266-2271.	0.7	33
52	Brain CT scan for pediatric minor accidental head injury. An Italian experience and review of literature. <i>Child's Nervous System</i> , 2012, 28, 1063-1068.	0.6	25
53	A multicentre retrospective study of 66 Italian children with food proteinâ€“induced enterocolitis syndrome: different management for different phenotypes. <i>Clinical and Experimental Allergy</i> , 2012, 42, 1257-1265.	1.4	179
54	Congenital syphilis: remember to not forget. <i>BMJ Case Reports</i> , 2012, 2012, .	0.2	2

#	ARTICLE	IF	CITATIONS
55	Isolated contact urticaria caused by immunoglobulin E-mediated fish allergy. <i>Israel Medical Association Journal</i> , 2012, 14, 11-3.	0.1	21
56	Pott's puffy tumour by <i>Streptococcus intermedius</i> a rare complication of sinusitis. <i>BMJ Case Reports</i> , 2011, 2011, bcr0820114660-bcr0820114660.	0.2	5
57	Is heel prick as safe as we think?. <i>BMJ Case Reports</i> , 2011, 2011, bcr0820114677-bcr0820114677.	0.2	7
58	Incomplete Kawasaki syndrome followed by systemic onset-juvenile idiopathic arthritis mimicking Kawasaki syndrome. <i>Rheumatology International</i> , 2010, 30, 535-539.	1.5	23
59	Responsiveness to intravenous immunoglobulins and occurrence of coronary artery abnormalities in a single-center cohort of Italian patients with Kawasaki syndrome. <i>Rheumatology International</i> , 2010, 30, 841-846.	1.5	57
60	Measles-induced respiratory distress, air-leak and ARDS. <i>European Journal of Clinical Microbiology and Infectious Diseases</i> , 2010, 29, 181-185.	1.3	14
61	Specific oral tolerance induction (SOTI) in pediatric age: Clinical research or just routine practice?. <i>Pediatric Allergy and Immunology</i> , 2010, 21, e446-9.	1.1	22
62	Intravenous Immunoglobulin therapy for anti-E hemolytic disease in the newborn. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2010, 23, 1059-1061.	0.7	5
63	Montelukast versus inhaled corticosteroids as monotherapy for prevention of asthma: which one is best?. <i>Allergologia Et Immunopathologia</i> , 2009, 37, 26-30.	1.0	6
64	The influence of quality criteria on parents' evaluation of medical web-pages: An Italian randomised trial. <i>Technology and Health Care</i> , 2007, 15, 399-406.	0.5	1
65	A quality evaluation methodology of health web-pages for non-professionals. <i>Informatics for Health and Social Care</i> , 2004, 29, 95-107.	1.0	19
66	The Evolution of Web-based Medical Information on Sore Throat: a Longitudinal Study. <i>Journal of Medical Internet Research</i> , 2003, 5, e10.	2.1	5