Luisa De Sanctis

List of Publications by Year
in descending order

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46 papers	2,335 citations	346980 22 h-index	263392 45 g-index
46 all docs	46 docs citations	46 times ranked	2550 citing authors

#	Article	IF	Citations
1	Hypogonadism in male and female: which is the best treatment?. Minerva Pediatrics, 2022, 73, .	0.2	1
2	Differences of sex development in the newborn: from clinical scenario to molecular diagnosis. Minerva Pediatrics, 2022, 73, .	0.2	5
3	The Hyperphagia Questionnaire: Insights From a Multicentric Validation Study in Individuals With Prader Willi Syndrome. Frontiers in Pediatrics, 2022, 10, 829486.	0.9	2
4	Mediterranean Dietary Treatment in Hyperlipidemic Children: Should It Be an Option?. Nutrients, 2022, 14, 1344.	1.7	4
5	Congenital Hypothyroidism: A 2020â€“2021 Consensus Guidelines Updateâ€”An ENDO-European Reference Network Initiative Endorsed by the European Society for Pediatric Endocrinology and the European Society for Endocrinology. Thyroid, 2021, 31, 387-419.	2.4	209
6	Inactivating PTH/PTHrP signaling disorders (iPPSDs): evaluation of the new classification in a multicenter large series of 544 molecularly characterized patients. European Journal of Endocrinology, 2021, 184, 311-320.	1.9	13
7	â€œPrimary Hyperparathyroidism (PHPT) in Children: Two Case Reports and Review of the Literatureâ€. Case Reports in Endocrinology, 2021, 2021, 1-6.	0.2	3
8	Multidisciplinary Approach for Hypothalamic Obesity in Children and Adolescents: A Preliminary Study. Children, 2021, 8, 531.	0.6	3
9	Biological clock and heredity in pubertal timing: what is new?. Minerva Pediatrics, 2021, , .	0.2	2
10	Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients. Hormone Research in Paediatrics, 2020, 93, 182-196.	0.8	42
11	Jaffe-Campanacci syndrome or neurofibromatosis type 1: a case report of phenotypic overlap with detection of NF1 gene mutation in non-ossifying fibroma. Italian Journal of Pediatrics, 2020, 46, 58.	1.0	6
12	Pretreatment Endocrine Disorders Due to Optic Pathway Gliomas in Pediatric Neurofibromatosis Type 1: Multicenter Study. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2214-e2221.	1.8	19
13	Prospective evaluation of autoimmune and non-autoimmune subclinical hypothyroidism in Down syndrome children. European Journal of Endocrinology, 2020, 182, 385-392.	1.9	17
14	Improved Molecular Diagnosis of McCuneâ€“Albright Syndrome and Bone Fibrous Dysplasia by Digital PCR. Frontiers in Genetics, 2019, 10, 862.	1.1	11
15	2q37 Deletions in Patients With an Albright Hereditary Osteodystrophy Phenotype and PTH Resistance. Frontiers in Endocrinology, 2019, 10, 604.	1.5	4
16	Growth hormoneâ€”Insulinâ€like growth factor 1 axis hyperactivity on bone fibrous dysplasia in McCuneâ€Albright Syndrome. Clinical Endocrinology, 2018, 89, 56-64.	1.2	21
17	Genetic and Epigenetic Defects at the GNAS Locus Lead to Distinct Patterns of Skeletal Growth but Similar Early-Onset Obesity. Journal of Bone and Mineral Research, 2018, 33, 1480-1488.	3.1	41
18	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	4.3	224

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19	Combining Real-Time COLD- and MAMA-PCR TaqMan Techniques to Detect and Quantify R201 <i>GNAS</i> Mutations in the McCune-Albright Syndrome. Hormone Research in Paediatrics, 2017, 87, 342-349.	0.8	14
20	Compound heterozygosity for two GHR missense mutations in a patient affected by Laron Syndrome: a case report. Italian Journal of Pediatrics, 2017, 43, 94.	1.0	5
21	Screening of PRKAR1A and PDE4D in a Large Italian Series of Patients Clinically Diagnosed With Albright Hereditary Osteodystrophy and/or Pseudohypoparathyroidism. Journal of Bone and Mineral Research, 2016, 31, 1215-1224.	3.1	47
22	Novel Microdeletions Affecting the GNAS Locus in Pseudohypoparathyroidism: Characterization of the Underlying Mechanisms. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E681-E687.	1.8	23
23	Iatrogenic acute pancreatitis due to hypercalcemia in a child with pseudohypoparathyroidism. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 149-52.	0.4	3
24	Quantitative Analysis of Methylation Defects and Correlation With Clinical Characteristics in Patients With Pseudohypoparathyroidism Type I and GNAS Epigenetic Alterations. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E508-E517.	1.8	49
25	Autosomal Dominant Pseudohypoparathyroidism Type Ib: A Novel Inherited Deletion AblatingSTX16Causes Loss of Imprinting at the A/B DMR. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E724-E728.	1.8	68
26	Fetal alcohol syndrome: new perspectives for an ancient and underestimated problem. Journal of Maternal-Fetal and Neonatal Medicine, 2011, 24, 34-37.	0.7	28
27	Neonatal hepatoblastoma in a newborn with severe phenotype of Beckwithâ€“Wiedemann syndrome. European Journal of Pediatrics, 2011, 170, 1407-1411.	1.3	38
28	Functional characterization of GNAS mutations found in patients with pseudohypoparathyroidism type Ic defines a new subgroup of pseudohypoparathyroidism affecting selectively Gsî±-receptor interaction. Human Mutation, 2011, 32, 653-660.	1.1	62
29	Recombinant Human GH Replacement Therapy in Children with Pseudohypoparathyroidism Type Ia: First Study on the Effect on Growth. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 5011-5017.	1.8	55
30	Pseudohypoparathyroidism andGNASEpigenetic Defects: Clinical Evaluation of Albright Hereditary Osteodystrophy and Molecular Analysis in 40 Patients. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 651-658.	1.8	144
31	Regulation of spermatogenesis in McCuneâ€“Albright syndrome: lessons from a 15-year follow-up.. European Journal of Endocrinology, 2008, 158, 921-927.	1.9	21
32	Elucidating the Underlying Molecular Pathogenesis ofNR3C2Mutants Causing Autosomal Dominant Pseudohypoaldosteronism Type 1. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 4552-4561.	1.8	58
33	McCune-Albright Syndrome in a Boy May Present with a Monolateral Macroorchidism as an Early and Isolated Clinical Manifestation. Hormone Research in Paediatrics, 2006, 65, 114-119.	0.8	19
34	Pseudohypoparathyroidism: History of the Disease. Journal of Pediatric Endocrinology and Metabolism, 2006, 19, 627-33.	0.4	3
35	SOX2anophthalmia syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 1-7.	0.7	194
36	Adult Height in Patients Treated for Isolated Growth Hormone Deficiency: Role of Birth Weight. Hormone Research in Paediatrics, 2005, 63, 102-106.	0.8	4

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37	Parental Origin of GsÎ± Mutations in the McCune-Albright Syndrome and in Isolated Endocrine Tumors. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 3007-3009.	1.8	61
38	Brachydactyly in 14 Genetically Characterized Pseudohypoparathyroidism Type Ia Patients. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 1650-1655.	1.8	56
39	Familial PAX8 Small Deletion (c.989_992delACCC) Associated with Extreme Phenotype Variability. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 5669-5674.	1.8	60
40	Dihydropteridine reductase deficiency in man: From biology to treatment. Medicinal Research Reviews, 2004, 24, 127-150.	5.0	76
41	Molecular Analysis of the GNAS1 Gene for the Correct Diagnosis of Albright Hereditary Osteodystrophy and Pseudohypoparathyroidism. Pediatric Research, 2003, 53, 749-755.	1.1	57
42	Comparison between SLC3A1 and SLC7A9 Cystinuria Patients and Carriers: A Need for a New Classification. Journal of the American Society of Nephrology: JASN, 2002, 13, 2547-2553.	3.0	234
43	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. Nature Genetics, 1999, 23, 52-57.	9.4	280
44	Molecular analysis of the cystinuria disease gene: identification of four new mutations, one large deletion, and one polymorphism. Human Genetics, 1996, 98, 447-451.	1.8	44
45	Novel missense mutation in the phenylalanine hydroxylase gene leading to complete loss of enzymatic activity. Human Mutation, 1995, 6, 247-249.	1.1	4
46	â€œLymphocyte population in peripheral blood in children and adolescents with graves disease. Potential predictive tool for severity of the diseaseâ€• Endocrine, 0, , .	1.1	1