

# Antonio Balsamo

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

113 papers	3,284 citations	32 h-index	54 g-index
116 ext. papers	3,786 ext. citations	4.2 avg, IF	4.33 L-index

#	Paper	IF	Citations
113	Differences of sex development in the newborn: from clinical scenario to molecular diagnosis. <i>Minerva Pediatrics</i> , <b>2021</b> ,	1.5	2
112	International practice of corticosteroid replacement therapy in congenital adrenal hyperplasia: data from the I-CAH registry. <i>European Journal of Endocrinology</i> , <b>2021</b> , 184, 553-563	6.5	4
111	Gonadectomy in conditions affecting sex development: a registry-based cohort study. <i>European Journal of Endocrinology</i> , <b>2021</b> , 184, 791-801	6.5	1
110	Real-World Estimates of Adrenal Insufficiency-Related Adverse Events in Children With Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2021</b> , 106, e192-e203	5.6	8
109	Primary Adrenal Insufficiency in Childhood: Data From a Large Nationwide Cohort. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2021</b> , 106, 762-773	5.6	5
108	Testosterone Therapy and Its Monitoring in Adolescent Boys with Hypogonadism: Results of an International Survey from the I-DSD Registry. <i>Sexual Development</i> , <b>2021</b> , 15, 236-243	1.6	1
107	Good overall behavioural adjustment in children and adolescents with classic congenital adrenal hyperplasia. <i>Endocrine</i> , <b>2020</b> , 68, 427-437	4	5
106	Congenital Adrenal Hyperplasias Presenting in the Newborn and Young Infant. <i>Frontiers in Pediatrics</i> , <b>2020</b> , 8, 593315	3.4	5
105	Growth Trajectory and Adult Height in Children with Nonclassical Congenital Adrenal Hyperplasia. <i>Hormone Research in Paediatrics</i> , <b>2020</b> , 93, 173-181	3.3	4
104	Plasma Renin Measurements are Unrelated to Mineralocorticoid Replacement Dose in Patients With Primary Adrenal Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2020</b> , 105,	5.6	15
103	Clinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2019</b> , 104, 4366-4381	5.6	14
102	46,XX DSD due to Androgen Excess in Monogenic Disorders of Steroidogenesis: Genetic, Biochemical, and Clinical Features. <i>International Journal of Molecular Sciences</i> , <b>2019</b> , 20,	6.3	12
101	Thyroid function in patients with Prader-Willi syndrome: an Italian multicenter study of 339 patients. <i>Journal of Pediatric Endocrinology and Metabolism</i> , <b>2019</b> , 32, 159-165	1.6	18
100	X-linked hypophosphatemic rickets: an Italian expertsSopinion survey. <i>Italian Journal of Pediatrics</i> , <b>2019</b> , 45, 67	3.2	20
99	Addressing gaps in care of people with conditions affecting sex development and maturation. <i>Nature Reviews Endocrinology</i> , <b>2019</b> , 15, 615-622	15.2	17
98	Novel non-classic CYP21A2 variants, including combined alleles, identified in patients with congenital adrenal hyperplasia. <i>Clinical Biochemistry</i> , <b>2019</b> , 73, 50-56	3.5	2
97	Familial neurohypophyseal diabetes insipidus in 13 kindreds and 2 novel mutations in the vasopressin gene. <i>European Journal of Endocrinology</i> , <b>2019</b> , 181, 233-244	6.5	8

96	Response to Letter to the Editor: "Clinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosis". <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2019</b> , 104, 5812-5813	5.6	
95	Normative Basal Values of Hormones and Proteins of Gonadal and Adrenal Functions from Birth to Adulthood. <i>Sexual Development</i> , <b>2018</b> , 12, 50-94	1.6	11
94	A genetic epidemiology study of congenital adrenal hyperplasia in Italy. <i>Clinical Genetics</i> , <b>2018</b> , 93, 223-227	5	
93	Characteristics of a nationwide cohort of patients presenting with isolated hypogonadotropic hypogonadism (IHH). <i>European Journal of Endocrinology</i> , <b>2018</b> , 178, 23-32	6.5	54
92	Diagnosis, treatment and prevention of pediatric obesity: consensus position statement of the Italian Society for Pediatric Endocrinology and Diabetology and the Italian Society of Pediatrics. <i>Italian Journal of Pediatrics</i> , <b>2018</b> , 44, 88	3.2	71
91	Mutational and functional studies on NR5A1 gene in 46,XY disorders of sex development: identification of six novel loss of function mutations. <i>Fertility and Sterility</i> , <b>2018</b> , 109, 1105-1113	4.8	9
90	Disorders of Sexual Development in Newborns <b>2018</b> , 1-25		
89	Disorders of Sexual Development in Newborns <b>2018</b> , 1893-1917		1
88	Endocrine Diseases and Disorders of Thyroid Function in Newborns <b>2018</b> , 1833-1891		
87	Endocrine Diseases and Disorders of Thyroid Function in Newborns <b>2018</b> , 1-62		
86	Evaluation of DSD training schools organized by cost action BM1303 "DSDnet". <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 227	4.2	2
85	Management of Gonads in Adults with Androgen Insensitivity: An International Survey. <i>Hormone Research in Paediatrics</i> , <b>2018</b> , 90, 236-246	3.3	18
84	Involving Individuals with Disorders of Sex Development and Their Parents in Exploring New Models of Shared Learning: Proceedings from a DSDnet COST Action Workshop. <i>Sexual Development</i> , <b>2018</b> ,	1.6	9
83	Childhood obesity classification systems and cardiometabolic risk factors: a comparison of the Italian, World Health Organization and International Obesity Task Force references. <i>Italian Journal of Pediatrics</i> , <b>2017</b> , 43, 19	3.2	26
82	Global Application of the Assessment of Communication Skills of Paediatric Endocrinology Fellows in the Management of Differences in Sex Development Using the ESPE E-Learning.Org Portal. <i>Hormone Research in Paediatrics</i> , <b>2017</b> , 88, 127-139	3.3	6
81	Bone Mineral Density in Women Living with Complete Androgen Insensitivity Syndrome and Intact Testes or Removed Gonads. <i>Sexual Development</i> , <b>2017</b> , 11, 182-189	1.6	19
80	Two Moroccan Sisters Presenting with a Severe Salt-Wasting Form of Congenital Adrenal Hyperplasia but Normal Female Genitalia. <i>Sexual Development</i> , <b>2017</b> , 11, 82-85	1.6	3
79	The rehabilitation of children and adolescents with severe or medically complicated obesity: an ISPED expert opinion document. <i>Eating and Weight Disorders</i> , <b>2017</b> , 22, 3-12	3.6	1

78	Improving the diagnosis of 11 $\beta$ -hydroxylase deficiency using home-made MLPA probes: identification of a novel chimeric CYP11B2/CYP11B1 gene in a Sicilian patient. <i>Journal of Endocrinological Investigation</i> , <b>2016</b> , 39, 291-5	5.2	2
77	Triglycerides-to-HDL cholesterol ratio as screening tool for impaired glucose tolerance in obese children and adolescents. <i>Acta Diabetologica</i> , <b>2016</b> , 53, 493-8	3.9	18
76	Growing Up with Type 1 Narcolepsy: Its Anthropometric and Endocrine Features. <i>Journal of Clinical Sleep Medicine</i> , <b>2016</b> , 12, 1649-1657	3.1	38
75	Disorders of Sexual Development in Newborns <b>2016</b> , 1-24		1
74	Endocrine Diseases and Disorders of Thyroid Function in Newborns <b>2016</b> , 1-60		
73	Quality of Life and Psychological Adjustment of Women Living with 46,XY Differences of Sex Development. <i>Journal of Sexual Medicine</i> , <b>2015</b> , 12, 1440-9	1.1	35
72	Novel associations in disorders of sex development: findings from the I-DSD Registry. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2014</b> , 99, E348-55	5.6	64
71	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency: functional consequences of four CYP11B1 mutations. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22, 610-6	5.3	33
70	Changes over time in sex assignment for disorders of sex development. <i>Pediatrics</i> , <b>2014</b> , 134, e710-5	7.4	81
69	Comment on "complete androgen insensitivity syndrome: optimizing diagnosis and management". <i>Case Reports in Obstetrics and Gynecology</i> , <b>2014</b> , 2014, 285715	0.8	2
68	Rare diseases research and practice. <i>Endocrine Development</i> , <b>2014</b> , 27, 234-56		8
67	Hirsutism and Virilization <b>2014</b> , 145-157		
66	Comparison between liquid and tablet formulations of levothyroxine in the initial treatment of congenital hypothyroidism. <i>Journal of Pediatrics</i> , <b>2013</b> , 162, 1264-9, 1269.e1-2	3.6	45
65	High prevalence of precocious puberty and obesity in childhood narcolepsy with cataplexy. <i>Sleep</i> , <b>2013</b> , 36, 175-81	1.1	88
64	Severe obesity and cardiometabolic risk in children: comparison from two international classification systems. <i>PLoS ONE</i> , <b>2013</b> , 8, e83793	3.7	17
63	Increased large artery intima media thickness in adolescents with either classical or non-classical congenital adrenal hyperplasia. <i>Journal of Endocrinological Investigation</i> , <b>2013</b> , 36, 12-5	5.2	10
62	Disorders of Sexual Development <b>2012</b> , 1004-1017		
61	A sequence variation in 3'UTR of CYP21A2 gene correlates with a mild form of congenital adrenal hyperplasia. <i>Journal of Endocrinological Investigation</i> , <b>2012</b> , 35, 298-305	5.2	10

60 Endocrine Diseases of Newborn **2012**, 967-992

59	Impact of molecular genetics on congenital adrenal hyperplasia management. <i>Sexual Development</i> , <b>2010</b> , 4, 233-48	1.6	42
58	Insulin resistance is a risk factor for high blood pressure regardless of body size and fat distribution in obese children. <i>Nutrition, Metabolism and Cardiovascular Diseases</i> , <b>2010</b> , 20, 266-73	4.5	24
57	Prevalence of pathogenetic MC4R mutations in Italian children with early onset obesity, tall stature and familial history of obesity. <i>BMC Medical Genetics</i> , <b>2009</b> , 10, 25	2.1	19
56	Characterization of deletions at 9p affecting the candidate regions for sex reversal and deletion 9p syndrome by MLPA. <i>European Journal of Human Genetics</i> , <b>2009</b> , 17, 1439-47	5.3	68
55	Gene dosage imbalances in patients with 46,XY gonadal DSD detected by an in-house-designed synthetic probe set for multiplex ligation-dependent probe amplification analysis. <i>Clinical Genetics</i> , <b>2008</b> , 73, 453-64	4	34
54	Three novel AMH gene mutations in a patient with persistent mullerian duct syndrome and normal AMH serum dosage. <i>Hormone Research</i> , <b>2008</b> , 70, 124-8		12
53	The role of 21-hydroxylase in the pathogenesis of adrenal masses: review of the literature and focus on our own experience. <i>Journal of Endocrinological Investigation</i> , <b>2007</b> , 30, 615-23	5.2	15
52	Emotion recognition and expression in young obese participants: preliminary study. <i>Perceptual and Motor Skills</i> , <b>2007</b> , 105, 477-82	2.2	17
51	Functional characterization of naturally occurring NR3C2 gene mutations in Italian patients suffering from pseudohypoaldosteronism type 1. <i>European Journal of Endocrinology</i> , <b>2007</b> , 156, 249-56	6.5	20
50	EMOTION RECOGNITION AND EXPRESSION IN YOUNG OBESE PARTICIPANTS: PRELIMINARY STUDY. <i>Perceptual and Motor Skills</i> , <b>2007</b> , 105, 477	2.2	2
49	Functional studies of two novel and two rare mutations in the 21-hydroxylase gene. <i>Journal of Molecular Medicine</i> , <b>2006</b> , 84, 521-8	5.5	25
48	Reproductive outcome in patients treated and not treated for idiopathic early puberty: long-term results of a randomized trial in adults. <i>Journal of Pediatrics</i> , <b>2006</b> , 149, 532-6	3.6	25
47	Effect on adult height of pubertal growth hormone retesting and withdrawal of therapy in patients with previously diagnosed growth hormone deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2006</b> , 91, 4271-6	5.6	35
46	Two novel GnRHR gene mutations in two siblings with hypogonadotropic hypogonadism. <i>European Journal of Endocrinology</i> , <b>2006</b> , 155, 201-5	6.5	19
45	Italian cross-sectional growth charts for height, weight and BMI (2 to 20 yr). <i>Journal of Endocrinological Investigation</i> , <b>2006</b> , 29, 581-93	5.2	559
44	Birth length and weight in congenital adrenal hyperplasia according to the different phenotypes. <i>European Journal of Pediatrics</i> , <b>2006</b> , 165, 380-3	4.1	13
43	SRD5A2 gene analysis in an Italian population of under-masculinized 46,XY subjects. <i>Clinical Endocrinology</i> , <b>2005</b> , 63, 375-80	3.4	45

42	A new DAX1 gene mutation associated with congenital adrenal hypoplasia and hypogonadotropic hypogonadism. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 135, 292-6	2.5	9
41	Functional analysis of two recurrent amino acid substitutions in the CYP21 gene from Italian patients with congenital adrenal hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 2402-7	5.6	45
40	CYP21 genotype, adult height, and pubertal development in 55 patients treated for 21-hydroxylase deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2003</b> , 88, 5680-8	5.6	61
39	Treatment for congenital hypothyroidism: thyroxine alone or thyroxine plus triiodothyronine?. <i>Pediatrics</i> , <b>2003</b> , 111, 1055-60	7.4	58
38	Deficit in the discrimination of nonverbal emotions in children with obesity and their mothers. <i>International Journal of Obesity</i> , <b>2003</b> , 27, 191-5	5.5	44
37	Italian cross-sectional growth charts for height, weight and BMI (6-20 y). <i>European Journal of Clinical Nutrition</i> , <b>2002</b> , 56, 171-80	5.2	230
36	Final height of short subjects of low birth weight with and without growth hormone treatment. <i>Archives of Disease in Childhood</i> , <b>2001</b> , 84, 340-3	2.2	32
35	CYP21 analysis and phenotype/genotype relationship in the screened population of the Italian Emilia-Romagna region. <i>Clinical Endocrinology</i> , <b>2000</b> , 53, 117-25	3.4	48
34	Randomised trial of LHRH analogue treatment on final height in girls with onset of puberty aged 7.5-8.5 years. <i>Archives of Disease in Childhood</i> , <b>1999</b> , 81, 329-32	2.2	87
33	Birth weight affects final height in patients treated for growth hormone deficiency. <i>Clinical Endocrinology</i> , <b>1999</b> , 51, 733-9	3.4	11
32	Involvement of the skull base and vault in chronic idiopathic hyperphosphatasia. <i>Pediatric Radiology</i> , <b>1999</b> , 29, 16-8	2.8	4
31	Low growth hormone-binding protein in infants with congenital hypothyroidism. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1998</b> , 83, 3643-6	5.6	15
30	Final height of patients treated for isolated GH deficiency: examination of 83 patients. <i>European Journal of Endocrinology</i> , <b>1997</b> , 137, 53-60	6.5	29
29	Decoding difficulties of facial expression of emotions in mothers of children suffering from developmental obesity. <i>Psychotherapy and Psychosomatics</i> , <b>1996</b> , 65, 258-61	9.4	20
28	Response to growth hormone therapy in patients with growth hormone deficiency who at birth were small or appropriate in size for gestational age. <i>Journal of Pediatrics</i> , <b>1995</b> , 126, 474-7	3.6	16
27	Empty sella in children and adolescents with possible hypothalamic-pituitary disorders. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1994</b> , 78, 767-71	5.6	52
26	Long-term follow-up and final height in girls with central precocious puberty treated with luteinizing hormone-releasing hormone analogue nasal spray. <i>JAMA Pediatrics</i> , <b>1994</b> , 148, 1194-9		23
25	Two mutations causing complete androgen insensitivity: a frame-shift in the steroid binding domain and a Cys->Phe substitution in the second zinc finger of the androgen receptor. <i>Human Molecular Genetics</i> , <b>1994</b> , 3, 1169-70	5.6	14

24	Molecular study of human growth hormone gene cluster in three families with isolated growth hormone deficiency and similar phenotype. <i>European Journal of Pediatrics</i> , <b>1994</b> , 153, 635-41	4.1	12
23	Empty sella in children and adolescents with possible hypothalamic- pituitary disorders. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1994</b> , 78, 767-771	5.6	43
22	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: Newborn screening and its relationship to the diagnosis and treatment of the disorder. <i>Screening: Journal of the International Society of Neonatal Screening</i> , <b>1993</b> , 2, 105-139		112
21	Pitfalls in diagnosing impaired growth hormone (GH) secretion: retesting after replacement therapy of 63 patients defined as GH deficient. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1992</b> , 74, 1284-9	5.6	84
20	Pitfalls in diagnosing impaired growth hormone (GH) secretion: retesting after replacement therapy of 63 patients defined as GH deficient. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1992</b> , 74, 1284-1289	5.6	60
19	Variability of growth hormone response to pharmacological and sleep tests performed twice in short children. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1990</b> , 71, 230-4	5.6	104
18	Influence of growth hormone on thymic endocrine activity in humans. <i>Hormone Research</i> , <b>1990</b> , 33, 248-55		50
17	Incidence of intratubular germ cell neoplasia in androgen insensitivity syndrome. <i>European Journal of Endocrinology</i> , <b>1990</b> , 123, 416-22	6.5	39
16	Long term growth hormone (GH)-releasing hormone and biosynthetic GH therapy in GH-deficient children: comparison of therapeutic effectiveness. <i>Journal of Endocrinological Investigation</i> , <b>1990</b> , 13, 235-9	5.2	3
15	Effect of gonadectomy on growth hormone, IGF-I and sex steroids in children with complete and incomplete androgen insensitivity. <i>European Journal of Endocrinology</i> , <b>1989</b> , 121, 777-83	6.5	20
14	Haemorheologic and fibrinolytic evaluation in obese children and adolescents. <i>European Journal of Pediatrics</i> , <b>1988</b> , 147, 381-4	4.1	11
13	Differential effects of 18- and 24-Gy cranial irradiation on growth rate and growth hormone release in children with prolonged survival after acute lymphocytic leukemia. <i>JAMA Pediatrics</i> , <b>1988</b> , 142, 1199-202		5
12	Neonatal screening programme for congenital adrenal hyperplasia in a homogeneous Caucasian population. <i>Journal of Inherited Metabolic Disease</i> , <b>1986</b> , 9 Suppl 1, 142-6	5.4	2
11	Decreased prolactin secretion in obesity. <i>Journal of Pediatrics</i> , <b>1986</b> , 109, 391	3.6	
10	Thyroid function and prolactin levels in insulin-dependent diabetic children and adolescents. <i>Diabetes</i> , <b>1984</b> , 33, 522-6	0.9	14
9	In vivo isotope study of the thyroid with <sup>99m</sup> TcO <sub>4</sub> <sup>-</sup> in neonatal congenital hypothyroidism. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , <b>1984</b> , 9, 351-3		4
8	How many cases of true precocious puberty in girls are idiopathic?. <i>Journal of Pediatrics</i> , <b>1983</b> , 102, 357-60		63
7	Neonatal screening for congenital adrenal hyperplasia using a microfilter paper method for 17-alpha-hydroxyprogesterone radioimmunoassay. Experience gained from the study of 22,233 cases. <i>Hormone Research</i> , <b>1982</b> , 16, 4-9		15



6	Hemostatic balance alterations in obese children. <i>Acta Paediatrica, International Journal of Paediatrics</i> , <b>1982</b> , 71, 479-84	3.1	4
5	Hamartomas of the tuber cinereum and precocious puberty. <i>Pediatric Neurosurgery</i> , <b>1982</b> , 9, 222-31	0.9	3
4	Neonatal screening for 21-hydroxylase deficiency: a microfilter paper method for 17-alpha-hydroxyprogesterone assay. <i>Journal of Endocrinological Investigation</i> , <b>1982</b> , 5, 87-90	5.2	13
3	Gonadal and adrenal secretion of dehydroepiandrosterone sulfate in prepubertal and pubertal subjects. <i>Journal of Endocrinological Investigation</i> , <b>1981</b> , 4, 197-202	5.2	5
2	Disordered prolactin secretion in the obese child and adolescent. <i>Archives of Disease in Childhood</i> , <b>1981</b> , 56, 386-9	2.2	14
1	Italian cross-sectional growth charts for height, weight and BMI (6-20 y)		3