

# Marco Bonomi

## List of Publications by Year in descending order

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110  
papers

3,692  
citations

156536

32  
h-index

156644

58  
g-index

117  
all docs

117  
docs citations

117  
times ranked

4237  
citing authors

#	ARTICLE	IF	CITATIONS
1	New and Consolidated Therapeutic Options for Pubertal Induction in Hypogonadism: In-depth Review of the Literature. <i>Endocrine Reviews</i> , 2022, 43, 824-851.	8.9	26
2	Pubertal induction and transition to adult sex hormone replacement in patients with congenital pituitary or gonadal reproductive hormone deficiency: an Endo-ERN clinical practice guideline. <i>European Journal of Endocrinology</i> , 2022, 186, G9-G49.	1.9	25
3	Prenatal dexamethasone treatment for classic 21-hydroxylase deficiency in Europe. <i>European Journal of Endocrinology</i> , 2022, 186, K17-K24.	1.9	7
4	First baseline data of the Klinefelter ItaliaN Group (KING) cohort: clinical features of adult with Klinefelter syndrome in Italy. <i>Journal of Endocrinological Investigation</i> , 2022, 45, 1769-1776.	1.8	1
5	Genetics of congenital hypogonadotropic hypogonadism: peculiarities and phenotype of an oligogenic disease. <i>Human Genetics</i> , 2021, 140, 77-111.	1.8	124
6	Central hypogonadism in Klinefelter syndrome: report of two cases and review of the literature. <i>Journal of Endocrinological Investigation</i> , 2021, 44, 459-470.	1.8	6
7	Clinical benefits of sex steroids given as a priming prior to GH provocative test or as a growth-promoting therapy in peripubertal growth delays: Results of a retrospective study among ENDO-ERN centres. <i>Clinical Endocrinology</i> , 2021, 94, 219-228.	1.2	12
8	ENDO-ERN expert opinion on the differential diagnosis of pubertal delay. <i>Endocrine</i> , 2021, 71, 681-688.	1.1	19
9	Hidden hypercortisolism: a too frequently neglected clinical condition. <i>Journal of Endocrinological Investigation</i> , 2021, 44, 1581-1596.	1.8	12
10	Cardiovascular risk and testosterone – from subclinical atherosclerosis to lipoprotein function to heart failure. <i>Reviews in Endocrine and Metabolic Disorders</i> , 2021, 22, 257-274.	2.6	26
11	Predictors of reproductive and non-reproductive outcomes of gonadotropin mediated pubertal induction in male patients with congenital hypogonadotropic hypogonadism (CHH). <i>Journal of Endocrinological Investigation</i> , 2021, 44, 2445-2454.	1.8	14
12	Thyrotropin Receptor p.N432D Retained Variant Is Degraded Through an Alternative Lysosomal/Autophagosomal Pathway and Can Be Functionally Rescued by Chemical Chaperones. <i>Thyroid</i> , 2021, 31, 1030-1040.	2.4	1
13	Pharmacological Induction of Puberty. , 2021, , .		2
14	Procoagulant Imbalance in Klinefelter Syndrome Assessed by Thrombin Generation Assay and Whole-Blood Thromboelastometry. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e1660-e1672.	1.8	7
15	Male and female sexual dysfunction in diabetic subjects: Focus on new antihyperglycemic drugs. <i>Reviews in Endocrine and Metabolic Disorders</i> , 2020, 21, 57-65.	2.6	24
16	Prokineticin receptor 2 affects GnRH3 neuron ontogeny but not fertility in zebrafish. <i>Scientific Reports</i> , 2020, 10, 7632.	1.6	4
17	Two novel truncating variants of the AAAS gene causative of the triple A syndrome. <i>Journal of Endocrinological Investigation</i> , 2020, 43, 973-982.	1.8	2
18	LGR4 deficiency results in delayed puberty through impaired Wnt/ $\beta$ 2-catenin signaling. <i>JCI Insight</i> , 2020, 5, .	2.3	25

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19	Mortality in an Italian nursing home during COVID-19 pandemic: correlation with gender, age, ADL, vitamin D supplementation, and limitations of the diagnostic tests. <i>Aging</i> , 2020, 12, 24522-24534.	1.4	56
20	Pubertal delay: the challenge of a timely differential diagnosis between congenital hypogonadotropic hypogonadism and constitutional delay of growth and puberty. <i>Minerva Pediatrica</i> , 2020, 72, 278-287.	2.6	9
21	Triple-A Syndrome (TAS): An In-Depth Overview on Genetic and Phenotype Heterogeneity. <i>Protein and Peptide Letters</i> , 2020, 27, 1192-1203.	0.4	6
22	Genetic and Epigenetic Aspects of the Supernumerary X Chromosome. <i>Trends in Andrology and Sexual Medicine</i> , 2020, , 25-35.	0.1	0
23	MON-273 Retrospective Analysis of Gonadotropin-Mediated Pubertal Induction in Male Patients with Congenital Hypogonadotropic Hypogonadism (CHH). <i>Journal of the Endocrine Society</i> , 2020, 4, .	0.1	0
24	Addressing gaps in care of people with conditions affecting sex development and maturation. <i>Nature Reviews Endocrinology</i> , 2019, 15, 615-622.	4.3	30
25	45,X/46,X,i(Yp): Importance of Assessment and Support during Puberty and Adolescence. <i>Sexual Development</i> , 2019, 13, 118-124.	1.1	2
26	Evidence for a Common Genetic Origin of Classic and Milder Adult-Onset Forms of Isolated Hypogonadotropic Hypogonadism. <i>Journal of Clinical Medicine</i> , 2019, 8, 126.	1.0	32
27	High-Density Lipoprotein Function Is Reduced in Patients Affected by Genetic or Idiopathic Hypogonadism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 3097-3107.	1.8	21
28	Selenium supplementation in the management of thyroid autoimmunity during pregnancy: results of the "SERENA study", a randomized, double-blind, placebo-controlled trial. <i>Endocrine</i> , 2019, 66, 542-550.	1.1	44
29	Thyroid function in Klinefelter syndrome: a multicentre study from KING group. <i>Journal of Endocrinological Investigation</i> , 2019, 42, 1199-1204.	1.8	15
30	The diagnosis and management of central hypothyroidism in 2018. <i>Endocrine Connections</i> , 2019, 8, R44-R54.	0.8	35
31	Neuropsychiatric Aspects in Men with Klinefelter Syndrome. <i>Endocrine, Metabolic and Immune Disorders - Drug Targets</i> , 2019, 19, 109-115.	0.6	20
32	A Rare SPRY4 Gene Mutation Is Associated With Anosmia and Adult-Onset Isolated Hypogonadotropic Hypogonadism. <i>Frontiers in Endocrinology</i> , 2019, 10, 781.	1.5	8
33	Genetics of nCHH: from a peculiar inheritance of a novel GNRHR mutation to a comprehensive review of the literature. <i>Andrology</i> , 2019, 7, 88-101.	1.9	11
34	Hypothalamus-Pituitary-Thyroid Axis. , 2019, , 398-402.		0
35	GnRH antagonist treatment of malignant adrenocortical tumors. <i>Endocrine-Related Cancer</i> , 2019, 26, 103-117.	1.6	14
36	OR06-5 Genetic Origin Of Classic And Milder Adult-onset Forms Of Isolated Hypogonadotropic Hypogonadism. <i>Journal of the Endocrine Society</i> , 2019, 3, .	0.1	0

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37	Clinical and genetic characterisation of a series of patients with triple A syndrome. European Journal of Pediatrics, 2018, 177, 363-369.	1.3	20
38	Characteristics of a nationwide cohort of patients presenting with isolated hypogonadotropic hypogonadism (IHH). European Journal of Endocrinology, 2018, 178, 23-32.	1.9	84
39	2018 European Thyroid Association (ETA) Guidelines on the Diagnosis and Management of Central Hypothyroidism. European Thyroid Journal, 2018, 7, 225-237.	1.2	135
40	A novel <i>IGSF1</i> mutation in a large Irish kindred highlights the need for familial screening in the <i>IGSF1</i> deficiency syndrome. Clinical Endocrinology, 2018, 89, 813-823.	1.2	16
41	Zebrafish Model for Investigating the Integrated Control of Reproduction. , 2018, , 323-333.		0
42	Hypogonadotropic hypogonadism and pituitary hypoplasia as recurrent features in Ulnar-Mammary syndrome. Endocrine Connections, 2018, 7, 1432-1441.	0.8	9
43	The multiple genetic causes of central hypothyroidism. Best Practice and Research in Clinical Endocrinology and Metabolism, 2017, 31, 255-263.	2.2	35
44	A frequent oligogenic involvement in congenital hypothyroidism. Human Molecular Genetics, 2017, 26, 2507-2514.	1.4	107
45	Mild <i>TSH</i> resistance: Clinical and hormonal features in childhood and adulthood. Clinical Endocrinology, 2017, 87, 587-596.	1.2	20
46	Control of GnRH Secretion. Endocrinology, 2017, , 3-33.	0.1	1
47	Developing and evaluating rare disease educational materials co-created by expert clinicians and patients: the paradigm of congenital hypogonadotropic hypogonadism. Orphanet Journal of Rare Diseases, 2017, 12, 57.	1.2	26
48	Genetics of primary ovarian insufficiency. Clinical Genetics, 2017, 91, 183-198.	1.0	155
49	Klinefelter syndrome (KS): genetics, clinical phenotype and hypogonadism. Journal of Endocrinological Investigation, 2017, 40, 123-134.	1.8	210
50	Switch to restoration therapy in a testosterone treated central hypogonadism with erythrocytosis. Endocrinology, Diabetes and Metabolism Case Reports, 2017, 2017, .	0.2	3
51	<i>IGSF1</i> Deficiency: Lessons From an Extensive Case Series and Recommendations for Clinical Management. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 1627-1636.	1.8	68
52	Control of GnRH Secretion. Endocrinology, 2016, , 1-31.	0.1	0
53	The zebrafish: an emerging animal model for investigating the hypothalamic regulation of reproduction. Minerva Endocrinologica, 2016, 41, 250-65.	1.7	9
54	The complex genetic basis of congenital hypogonadotropic hypogonadism. Minerva Endocrinologica, 2016, 41, 223-39.	1.7	16

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55	Congenital GnRH deficiency: a complex and genetically heterogeneous disease affecting human fertility and sexual development. <i>Minerva Endocrinologica</i> , 2016, 41, 183-7.	1.7	2
56	Central Hypothyroidism. , 2015, , 608-612.		0
57	A new variant in signal peptide of the human luteinizing hormone receptor (LHCGR) affects receptor biogenesis causing leydig cell hypoplasia. <i>Human Molecular Genetics</i> , 2015, 24, 6003-6012.	1.4	24
58	Central Hypothyroidism. , 2015, , 85-94.		0
59	Thyroid-Stimulating Hormone (TSH). , 2014, , .		0
60	Germline Prokineticin Receptor 2 (PROKR2) Variants Associated With Central Hypogonadism Cause Differential Modulation of Distinct Intracellular Pathways. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E458-E463.	1.8	21
61	Wasting syndrome with deep bradycardia as presenting manifestation of long-standing severe male hypogonadotropic hypogonadism: a case series. <i>BMC Endocrine Disorders</i> , 2014, 14, 78.	0.9	2
62	Uncertainties in endocrine substitution therapy for central endocrine insufficiencies. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , 2014, 124, 397-405.	1.0	5
63	Kallmann's syndrome and normosmic isolated hypogonadotropic hypogonadism: two largely overlapping manifestations of one rare disorder. <i>Journal of Endocrinological Investigation</i> , 2014, 37, 499-500.	1.8	8
64	Modern Methods to Investigate the Oligomerization of Glycoprotein Hormone Receptors (TSHR, LHR,) Tj ETQq0 0 0 rgBT /Overlock 10 T	0.4	4
65	The IGSF1 Deficiency Syndrome: Characteristics of Male and Female Patients. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, 4942-4952.	1.8	81
66	Loss-of-function mutations in the immunoglobulin superfamily member 1 gene (IGSF1) cause a novel, X-linked syndrome of central hypothyroidism and testicular enlargement. <i>Lancet, The</i> , 2013, 381, S15.	6.3	0
67	The unusual adequate development of a child with severe central hypothyroidism negative at neonatal thyrotropin screening. <i>Journal of Endocrinological Investigation</i> , 2013, 36, 788-9.	1.8	2
68	Frequent TSH Receptor Genetic Alterations with Variable Signaling Impairment in a Large Series of Children with Nonautoimmune Isolated Hyperthyrotropinemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E156-E160.	1.8	47
69	New understandings of the genetic basis of isolated idiopathic central hypogonadism. <i>Asian Journal of Andrology</i> , 2012, 14, 49-56.	0.8	74
70	Diagnostic and therapeutic challenges of acquired thyrotropic deficiency. <i>Annales D'Endocrinologie</i> , 2012, 73, 138-140.	0.6	4
71	Loss-of-function mutations in IGSF1 cause an X-linked syndrome of central hypothyroidism and testicular enlargement. <i>Nature Genetics</i> , 2012, 44, 1375-1381.	9.4	169
72	Increased loss of the Y chromosome in peripheral blood cells in male patients with autoimmune thyroiditis. <i>Journal of Autoimmunity</i> , 2012, 38, J193-J196.	3.0	64

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73	Blood Cell Mitochondrial DNA Content and Premature Ovarian Aging. PLoS ONE, 2012, 7, e42423.	1.1	37
74	Syndromes of resistance to TSH. Annales D'Endocrinologie, 2011, 72, 60-63.	0.6	9
75	Absence of primary hypothyroidism and goiter in Slc26a4 (-/-) mice fed on a low iodine diet. Journal of Endocrinological Investigation, 2011, 34, 593-8.	1.8	17
76	Rituximab treatment in patients with active Graves' orbitopathy: effects on proinflammatory and humoral immune reactions. Clinical and Experimental Immunology, 2010, 161, 436-443.	1.1	49
77	Genetics and phenomics of hypothyroidism due to TSH resistance. Molecular and Cellular Endocrinology, 2010, 322, 72-82.	1.6	87
78	Familial Glucocorticoid Deficiency (FGD) as a Cause of Hyperthyrotro-Pinemia (Hyper-TSH).. , 2010, , P3-325-P3-325.		0
79	A Family with Complete Resistance to Thyrotropin-Releasing Hormone. New England Journal of Medicine, 2009, 360, 731-734.	13.9	101
80	Primary ovarian insufficiency: X chromosome defects and autoimmunity. Journal of Autoimmunity, 2009, 33, 35-41.	3.0	100
81	Mitochondrial content in peripheral blood cells is reduced in women with compromised ovarian reserve. Fertility and Sterility, 2008, 90, S265.	0.5	0
82	Activator Protein-1 and Smad Proteins Synergistically Regulate Human Follicle-Stimulating Hormone $\beta$ 2-Promoter Activity. Endocrinology, 2008, 149, 5577-5591.	1.4	81
83	Technology Insight: modern methods to monitor proteinâ€“protein interactions reveal functional TSH receptor oligomerization. Nature Clinical Practice Endocrinology and Metabolism, 2007, 3, 180-190.	2.9	37
84	Syndromes of hormone resistance in the hypothalamicâ€“pituitaryâ€“thyroid axis. Best Practice and Research in Clinical Endocrinology and Metabolism, 2006, 20, 529-546.	2.2	66
85	Optimized Synthesis of AMPA Receptor Antagonist ZKâ€“187638 and Neurobehavioral Activity in a Mouse Model of Neuronal Ceroid Lipofuscinosis. ChemMedChem, 2006, 1, 1142-1148.	1.6	22
86	Structural Differences in the Hinge Region of the Glycoprotein Hormone Receptors: Evidence from the Sulfated Tyrosine Residues. Molecular Endocrinology, 2006, 20, 3351-3363.	3.7	79
87	Glycoprotein hormone receptors: link between receptor homodimerization and negative cooperativity. EMBO Journal, 2005, 24, 1954-1964.	3.5	266
88	Ipotiroidismo centrale: diagnosi, patogenesi e terapia sostitutiva. L Endocrinologo, 2005, 6, 89-96.	0.0	0
89	Delineation of the Discontinuous-Conformational Epitope of a Monoclonal Antibody Displaying Fullin Vitroand in VivoThyrotropin Activity. Molecular Endocrinology, 2004, 18, 3020-3034.	3.7	67
90	Modulation of Ligand Selectivity Associated with Activation of the Transmembrane Region of the Human Follitropin Receptor. Molecular Endocrinology, 2004, 18, 2061-2073.	3.7	81

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91	Glycoprotein Hormone Receptors: A Unique Paradigm for Ligand Binding and GPCR Activation. , 2003, , 161-166.		0
92	Highly Sensitive Serum Thyroglobulin and Circulating Thyroglobulin mRNA Evaluations in the Management of Patients with Differentiated Thyroid Cancer in Apparent Remission. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 3201-3208.	1.8	43
93	Generation of a mouse monoclonal TSH receptor antibody with stimulating activity. Biochemical and Biophysical Research Communications, 2002, 299, 891-896.	1.0	74
94	Tyrosine sulfation is required for agonist recognition by glycoprotein hormone receptors. EMBO Journal, 2002, 21, 504-513.	3.5	200
95	Decreased platelet glutamate uptake in patients with amyotrophic lateral sclerosis. Neurology, 2001, 56, 270-272.	1.5	67
96	Hyperplastic Pituitary Gland, High Serum Glycoprotein Hormone $\beta$ -Subunit, and Variable Circulating Thyrotropin (TSH) Levels as Hallmark of Central Hypothyroidism due to Mutations of the TSH $\beta$ Gene. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 1600-1604.	1.8	63
97	Hyperplastic Pituitary Gland, High Serum Glycoprotein Hormone $\beta$ -Subunit, and Variable Circulating Thyrotropin (TSH) Levels as Hallmark of Central Hypothyroidism due to Mutations of the TSH $\beta$ Gene. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 1600-1604.	1.8	69
98	Digenic and oligogenic cases in a large cohort of idiopathic central hypogonadism (ICH) patients. Endocrine Abstracts, 0, , .	0.0	0
99	Genotype and phenotype characterization of the cohort of Italian patients with idiopathic central hypogonadism (ICH). Endocrine Abstracts, 0, , .	0.0	0
100	Clinical and genetic findings of an Italian series of patients with ACTH resistance syndromes. Endocrine Abstracts, 0, , .	0.0	0
101	Zebrafish tool for the study of prokineticin receptor 2 (PROKR2) pathway on GNRH3 neuronal development. Endocrine Abstracts, 0, , .	0.0	0
102	Selenium supplementation in the management of thyroid autoimmunity during pregnancy: results of the 'Serena Study' a randomized, double-blind, placebo-controlled trial. Endocrine Abstracts, 0, , .	0.0	0
103	Knocking down/out the prokineticin pathway during zebrafish development results in the GnRH neurons axons misguiding. Endocrine Abstracts, 0, , .	0.0	0
104	Evaluation of genetic predisposition in severe and mild phenotypes of isolated hypogonadotropic hypogonadism. Endocrine Abstracts, 0, , .	0.0	0
105	Male patients with hypogonadism have an impaired lipoprotein function. Endocrine Abstracts, 0, , .	0.0	0
106	Characteristics, geographical distribution and age at diagnosis of patients with Klinefelter syndrome in Italy: a cohort study from the Klinefelter Italian Group (KING). Endocrine Abstracts, 0, , .	0.0	0
107	Clinical and genetic characterization of two cases of central hypogonadism in Klinefelter syndrome. Endocrine Abstracts, 0, , .	0.0	0
108	Coagulation abnormalities in patients with klinefelter syndrome compared to age-matched healthy controls: Cross-sectional assessment by thrombin generation test. Endocrine Abstracts, 0, , .	0.0	0

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109	Genetic background and previous androgenization are associated with reproductive and non-reproductive outcomes of Gonadotropin-mediated pubertal induction in Congenital Hypogonadotropic Hypogonadism (CHH). <i>Endocrine Abstracts</i> , 0, , .	0.0	1
110	Vitamin D and COVID-19 Severity and Related Mortality: A Prospective Study in Italy. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0