Marco Bonomi

List of Publications by Year in descending order

111 papers	3,692 citations	136950 32 h-index	138484 58 g-index
117 all docs	117 docs citations	117 times ranked	3951 citing authors

#	Article	IF	Citations
1	Glycoprotein hormone receptors: link between receptor homodimerization and negative cooperativity. EMBO Journal, 2005, 24, 1954-1964.	7.8	266
2	Klinefelter syndrome (KS): genetics, clinical phenotype and hypogonadism. Journal of Endocrinological Investigation, 2017, 40, 123-134.	3.3	210
3	Tyrosine sulfation is required for agonist recognition by glycoprotein hormone receptors. EMBO Journal, 2002, 21, 504-513.	7.8	200
4	Loss-of-function mutations in IGSF1 cause an X-linked syndrome of central hypothyroidism and testicular enlargement. Nature Genetics, 2012, 44, 1375-1381.	21.4	169
5	Genetics of primary ovarian insufficiency. Clinical Genetics, 2017, 91, 183-198.	2.0	155
6	2018 European Thyroid Association (ETA) Guidelines on the Diagnosis and Management of Central Hypothyroidism. European Thyroid Journal, 2018, 7, 225-237.	2.4	135
7	Genetics of congenital hypogonadotropic hypogonadism: peculiarities and phenotype of an oligogenic disease. Human Genetics, 2021, 140, 77-111.	3.8	124
8	A frequent oligogenic involvement in congenital hypothyroidism. Human Molecular Genetics, 2017, 26, 2507-2514.	2.9	107
9	A Family with Complete Resistance to Thyrotropin-Releasing Hormone. New England Journal of Medicine, 2009, 360, 731-734.	27.0	101
10	Primary ovarian insufficiency: X chromosome defects and autoimmunity. Journal of Autoimmunity, 2009, 33, 35-41.	6.5	100
11	Genetics and phenomics of hypothyroidism due to TSH resistance. Molecular and Cellular Endocrinology, 2010, 322, 72-82.	3.2	87
12	Characteristics of a nationwide cohort of patients presenting with isolated hypogonadotropic hypogonadism (IHH). European Journal of Endocrinology, 2018, 178, 23-32.	3.7	84
13	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
14	Activator Protein-1 and Smad Proteins Synergistically Regulate Human Follicle-Stimulating Hormone Î²-Promoter Activity. Endocrinology, 2008, 149, 5577-5591.	2.8	81
15	The IGSF1 Deficiency Syndrome: Characteristics of Male and Female Patients. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 4942-4952.	3.6	81
16	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
17	Generation of a mouse monoclonal TSH receptor antibody with stimulating activity. Biochemical and Biophysical Research Communications, 2002, 299, 891-896.	2.1	74
18	New understandings of the genetic basis of isolated idiopathic central hypogonadism. Asian Journal of Andrology, 2012, 14, 49-56.	1.6	74

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19	Hyperplastic Pituitary Gland, High Serum Glycoprotein Hormone Â-Subunit, and Variable Circulating Thyrotropin (TSH) Levels as Hallmark of Central Hypothyroidism due to Mutations of the TSHÂ Gene. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 1600-1604.	3.6	69
20	IGSF1 Deficiency: Lessons From an Extensive Case Series and Recommendations for Clinical Management. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 1627-1636.	3.6	68
21	Decreased platelet glutamate uptake in patients with amyotrophic lateral sclerosis. Neurology, 2001, 56, 270-272.	1.1	67
22	Delineation of the Discontinuous-Conformational Epitope of a Monoclonal Antibody Displaying Fullin Vitroandin VivoThyrotropin Activity. Molecular Endocrinology, 2004, 18, 3020-3034.	3.7	67
23	Syndromes of hormone resistance in the hypothalamicâ€“pituitaryâ€“thyroid axis. Best Practice and Research in Clinical Endocrinology and Metabolism, 2006, 20, 529-546.	4.7	66
24	Increased loss of the Y chromosome in peripheral blood cells in male patients with autoimmune thyroiditis. Journal of Autoimmunity, 2012, 38, J193-J196.	6.5	64
25	Hyperplastic Pituitary Gland, High Serum Glycoprotein HormoneÎ± -Subunit, and Variable Circulating Thyrotropin (TSH) Levels as Hallmark of Central Hypothyroidism due to Mutations of the TSHÎ² Gene¹. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 1600-1604.	3.6	63
26	Mortality in an Italian nursing home during COVID-19 pandemic: correlation with gender, age, ADL, vitamin D supplementation, and limitations of the diagnostic tests. Aging, 2020, 12, 24522-24534.	3.1	56
27	Rituximab treatment in patients with active Graves' orbitopathy: effects on proinflammatory and humoral immune reactions. Clinical and Experimental Immunology, 2010, 161, 436-443.	2.6	49
28	Frequent TSH Receptor Genetic Alterations with Variable Signaling Impairment in a Large Series of Children with Nonautoimmune Isolated Hyperthyrotropinemia. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E156-E160.	3.6	47
29	Selenium supplementation in the management of thyroid autoimmunity during pregnancy: results of the â€œSERENA studyâ€, a randomized, double-blind, placebo-controlled trial. Endocrine, 2019, 66, 542-550.	2.3	44
30	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.	3.6	43
31	Technology Insight: modern methods to monitor proteinâ€“protein interactions reveal functional TSH receptor oligomerization. Nature Clinical Practice Endocrinology and Metabolism, 2007, 3, 180-190.	2.8	37
32	Blood Cell Mitochondrial DNA Content and Premature Ovarian Aging. PLoS ONE, 2012, 7, e42423.	2.5	37
33	The multiple genetic causes of central hypothyroidism. Best Practice and Research in Clinical Endocrinology and Metabolism, 2017, 31, 255-263.	4.7	35
34	The diagnosis and management of central hypothyroidism in 2018. Endocrine Connections, 2019, 8, R44-R54.	1.9	35
35	Evidence for a Common Genetic Origin of Classic and Milder Adult-Onset Forms of Isolated Hypogonadotropic Hypogonadism. Journal of Clinical Medicine, 2019, 8, 126.	2.4	32
36	Addressing gaps in care of people with conditions affecting sex development and maturation. Nature Reviews Endocrinology, 2019, 15, 615-622.	9.6	30

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37	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.	2.7	26
38	Cardiovascular risk and testosterone â€“ from subclinical atherosclerosis to lipoprotein function to heart failure. Reviews in Endocrine and Metabolic Disorders, 2021, 22, 257-274.	5.7	26
39	New and Consolidated Therapeutic Options for Pubertal Induction in Hypogonadism: In-depth Review of the Literature. Endocrine Reviews, 2022, 43, 824-851.	20.1	26
40	LGR4 deficiency results in delayed puberty through impaired Wnt/Î²-catenin signaling. JCI Insight, 2020, 5, .	5.0	25
41	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. European Journal of Endocrinology, 2022, 186, G9-G49.	3.7	25
42	A new variant in signal peptide of the human luteinizing hormone receptor (LHCGR) affects receptor biogenesis causing leydig cell hypoplasia. Human Molecular Genetics, 2015, 24, 6003-6012.	2.9	24
43	Male and female sexual dysfunction in diabetic subjects: Focus on new antihyperglycemic drugs. Reviews in Endocrine and Metabolic Disorders, 2020, 21, 57-65.	5.7	24
44	Optimized Synthesis of AMPA Receptor Antagonist ZKâ€…187638 and Neurobehavioral Activity in a Mouse Model of Neuronal Ceroid Lipofuscinosis. ChemMedChem, 2006, 1, 1142-1148.	3.2	22
45	Germline Prokineticin Receptor 2 (PROKR2) Variants Associated With Central Hypogonadism Cause Differental Modulation of Distinct Intracellular Pathways. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E458-E463.	3.6	21
46	High-Density Lipoprotein Function Is Reduced in Patients Affected by Genetic or Idiopathic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3097-3107.	3.6	21
47	Mild <scp>TSH</scp> resistance: Clinical and hormonal features in childhood and adulthood. Clinical Endocrinology, 2017, 87, 587-596.	2.4	20
48	Clinical and genetic characterisation of a series of patients with triple A syndrome. European Journal of Pediatrics, 2018, 177, 363-369.	2.7	20
49	Neuropsychiatric Aspects in Men with Klinefelter Syndrome. Endocrine, Metabolic and Immune Disorders - Drug Targets, 2019, 19, 109-115.	1.2	20
50	ENDO-ERN expert opinion on the differential diagnosis of pubertal delay. Endocrine, 2021, 71, 681-688.	2.3	19
51	Absence of primary hypothyroidism and goiter in Slc26a4 (-/-) mice fed on a low iodine diet. Journal of Endocrinological Investigation, 2011, 34, 593-8.	3.3	17
52	A novel <scp>IGSF</scp>1 mutation in a large Irish kindred highlights the need for familial screening in the <scp>IGSF</scp>1 deficiency syndrome. Clinical Endocrinology, 2018, 89, 813-823.	2.4	16
53	The complex genetic basis of congenital hypogonadotropic hypogonadism. Minerva Endocrinologica, 2016, 41, 223-39.	1.8	16
54	Thyroid function in Klinefelter syndrome: a multicentre study from KING group. Journal of Endocrinological Investigation, 2019, 42, 1199-1204.	3.3	15

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55	Predictors of reproductive and non-reproductive outcomes of gonadotropin mediated pubertal induction in male patients with congenital hypogonadotropic hypogonadism (CHH). Journal of Endocrinological Investigation, 2021, 44, 2445-2454.	3.3	14
56	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.	3.6	14
57	GnRH antagonist treatment of malignant adrenocortical tumors. Endocrine-Related Cancer, 2019, 26, 103-117.	3.1	14
58	Clinical benefits of sex steroids given as a priming prior to GH provocative test or as a growthâ€promoting therapy in peripubertal growth delays: Results of a retrospective study among ENDOâ€ERN centres. Clinical Endocrinology, 2021, 94, 219-228.	2.4	12
59	Hidden hypercortisolism: a too frequently neglected clinical condition. Journal of Endocrinological Investigation, 2021, 44, 1581-1596.	3.3	12
60	Genetics of ncHH: from a peculiar inheritance of a novel GNRHR mutation to a comprehensive review of the literature. Andrology, 2019, 7, 88-101.	3.5	11
61	Syndromes of resistance to TSH. Annales D'Endocrinologie, 2011, 72, 60-63.	1.4	9
62	Hypogonadotropic hypogonadism and pituitary hypoplasia as recurrent features in Ulnar-Mammary syndrome. Endocrine Connections, 2018, 7, 1432-1441.	1.9	9
63	Pubertal delay: the challenge of a timely differential diagnosis between congenital hypogonadotropic hypogonadism and constitutional delay of growth and puberty. Minerva Pediatrica, 2020, 72, 278-287.	2.7	9
64	The zebrafish: an emerging animal model for investigating the hypothalamic regulation of reproduction. Minerva Endocrinologica, 2016, 41, 250-65.	1.8	9
65	Kallmannâ€™s syndrome and normosmic isolated hypogonadotropic hypogonadism: two largely overlapping manifestations of one rare disorder. Journal of Endocrinological Investigation, 2014, 37, 499-500.	3.3	8
66	A Rare SPRY4 Gene Mutation Is Associated With Anosmia and Adult-Onset Isolated Hypogonadotropic Hypogonadism. Frontiers in Endocrinology, 2019, 10, 781.	3.5	8
67	Procoagulant Imbalance in Klinefelter Syndrome Assessed by Thrombin Generation Assay and Whole-Blood Thromboelastometry. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e1660-e1672.	3.6	7
68	Prenatal dexamethasone treatment for classic 21-hydroxylase deficiency in Europe. European Journal of Endocrinology, 2022, 186, K17-K24.	3.7	7
69	Central hypogonadism in Klinefelter syndrome: report of two cases and review of the literature. Journal of Endocrinological Investigation, 2021, 44, 459-470.	3.3	6
70	Triple-A Syndrome (TAS): An In-Depth Overview on Genetic and Phenotype Heterogeneity. Protein and Peptide Letters, 2020, 27, 1192-1203.	0.9	6
71	Uncertainties in endocrine substitution therapy for central endocrine insufficiencies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2014, 124, 397-405.	1.8	5
72	Diagnostic and therapeutic challenges of acquired thyrotropic deficiency. Annales D'Endocrinologie, 2012, 73, 138-140.	1.4	4

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73	Modern Methods to Investigate the Oligomerization of Glycoprotein Hormone Receptors (TSHR, LHR,) Tj ETQq1 1 0.784314 rgBT /Ov	1.0	4
74	Prokineticin receptor 2 affects GnRH3 neuron ontogeny but not fertility in zebrafish. Scientific Reports, 2020, 10, 7632.	3.3	4
75	Switch to restoration therapy in a testosterone treated central hypogonadism with erythrocytosis. Endocrinology, Diabetes and Metabolism Case Reports, 2017, 2017, .	0.5	3
76	Wasting syndrome with deep bradycardia as presenting manifestation of long-standing severe male hypogonadotropic hypogonadism: a case series. BMC Endocrine Disorders, 2014, 14, 78.	2.2	2
77	45,X/46,X,i(Yp): Importance of Assessment and Support during Puberty and Adolescence. Sexual Development, 2019, 13, 118-124.	2.0	2
78	Two novel truncating variants of the AAAS gene causative of the triple A syndrome. Journal of Endocrinological Investigation, 2020, 43, 973-982.	3.3	2
79	Pharmacological Induction of Puberty. , 2021, , .		2
80	Congenital GnRH deficiency: a complex and genetically heterogeneous disease affecting human fertility and sexual development. Minerva Endocrinologica, 2016, 41, 183-7.	1.8	2
81	The unusual adequate development of a child with severe central hypothyroidsm negative at neonatal thyrotropin screening. Journal of Endocrinological Investigation, 2013, 36, 788-9.	3.3	2
82	Control of GnRH Secretion. Endocrinology, 2017, , 3-33.	0.1	1
83	Thyrotropin Receptor p.N432D Retained Variant Is Degraded Through an Alternative Lysosomal/Autophagosomal Pathway and Can Be Functionally Rescued by Chemical Chaperones. Thyroid, 2021, 31, 1030-1040.	4.5	1
84	Genetic background and previous androgenization are associated with reproductive and non-reproductive outcomes of Gonadotropin-mediated pubertal induction in Congenital Hypogonadotropic Hypogonadism (CHH). Endocrine Abstracts, 0, , .	0.0	1
85	First baseline data of the Klinefelter ItaliaN Group (KING) cohort: clinical features of adult with Klinefelter syndrome in Italy. Journal of Endocrinological Investigation, 2022, 45, 1769-1776.	3.3	1
86	Ipotiroidismo centrale: diagnosi, patogenesi e terapia sostitutiva. L Endocrinologo, 2005, 6, 89-96.	0.0	0
87	Mitochondrial content in peripheral blood cells is reduced in women with compromised ovarian reserve. Fertility and Sterility, 2008, 90, S265.	1.0	0
88	Loss-of-function mutations in the immunoglobulin superfamily member 1 gene (IGSF1) cause a novel, X-linked syndrome of central hypothyroidism and testicular enlargement. Lancet, The, 2013, 381, S15.	13.7	0
89	Thyroid-Stimulating Hormone (TSH). , 2014, , .		0
90	Central Hypothyroidism. , 2015, , 608-612.		0

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91	Central Hypothyroidism. , 2015, , 85-94.		0
92	Zebrafish Model for Investigating the Integrated Control of Reproduction. , 2018, , 323-333.		0
93	Hypothalamusâ€“Pituitaryâ€“Thyroid Axis. , 2019, , 398-402.		0
94	Glycoprotein Hormone Receptors: A Unique Paradigm for Ligand Binding and GPCR Activation. , 2003, , 161-166.		0
95	Familial Glucocorticoid Deficiency (FGD) as a Cause of Hyperthyrotro-Pinemia (Hyper-TSH).. , 2010, , P3-325-P3-325.		0
96	Digenic and oligogenic cases in a large cohort of idiopathic central hypogonadism (ICH) patients. Endocrine Abstracts, 0, , .	0.0	0
97	Genotype and phenotype characterization of the cohort of Italian patients with idiopathic central hypogonadism (ICH). Endocrine Abstracts, 0, , .	0.0	0
98	Clinical and genetic findings of an Italian series of patients with ACTH resistance syndromes. Endocrine Abstracts, 0, , .	0.0	0
99	Control of GnRH Secretion. Endocrinology, 2016, , 1-31.	0.1	0
100	Zebrafish tool for the study of prokineticin receptor 2 (PROKR2) pathway on GNRH3 neuronal development. Endocrine Abstracts, 0, , .	0.0	0
101	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
102	Knocking down/out the prokineticin pathway during zebrafish development results in the GnRH neurons axons misguiding. Endocrine Abstracts, 0, , .	0.0	0
103	Evaluation of genetic predisposition in severe and mild phenotypes of isolated hypogonadotropic hypogonadism. Endocrine Abstracts, 0, , .	0.0	0
104	OR06-5 Genetic Origin Of Classic And Milder Adult-onset Forms Of Isolated Hypogonadotropic Hypogonadism. Journal of the Endocrine Society, 2019, 3, .	0.2	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 and Epigenetic Aspects of the Supernumerary X Chromosome. Trends in Andrology and Sexual Medicine, 2020, , 25-35.	0.1	0
110	Vitamin D and COVID-19 Severity and Related Mortality: A Prospective Study in Italy. SSRN Electronic Journal, 0, , .	0.4	0
111	MON-273 Retrospective Analysis of Gonadotropin-Mediated Pubertal Induction in Male Patients with Congenital Hypogonadotropic Hypogonadism (CHH). Journal of the Endocrine Society, 2020, 4, .	0.2	0