Giovanna Weber

List of Publications by Year
in descending order

97 papers	4,492 citations	87843 38 h-index	114418 63 g-index
100 all docs	100 docs citations	100 times ranked	4085 citing authors

#	Article	IF	Citations
1	Neurosurgical treatment of craniopharyngioma in adults and children: early and long-term results in a large case series. Journal of Neurosurgery, 2011, 114, 1350-1359.	0.9	226
2	A 7â€year experience with low blood TSH cutoff levels for neonatal screening reveals an unsuspected frequency of congenital hypothyroidism (CH). Clinical Endocrinology, 2009, 71, 739-745.	1.2	207
3	Biallelic Inactivation of the Dual Oxidase Maturation Factor 2 (DUOXA2) Gene as a Novel Cause of Congenital Hypothyroidism. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 605-610.	1.8	157
4	Growth Hormone-Releasing Hormone Resistance in Pseudohypoparathyroidism Type Ia: New Evidence for Imprinting of the Gsî± Gene. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 4070-4074.	1.8	140
5	Accuracy of Fine Needle Aspiration Biopsy of Thyroid Nodules in Detecting Malignancy in Childhood: Comparison with Conventional Clinical, Laboratory, and Imaging Approaches. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 4644-4648.	1.8	139
6	Urinary Markers of Bone Turnover in Healthy Children and Adolescents: Age-Related Changes and Effect of Puberty. Calcified Tissue International, 1998, 63, 369-374.	1.5	136
7	Reversal of low bone density with a gluten-free diet in children and adolescents with celiac disease. American Journal of Clinical Nutrition, 1998, 67, 477-481.	2.2	129
8	Vitamin D in childhood and adolescence: an expert position statement. European Journal of Pediatrics, 2015, 174, 565-576.	1.3	129
9	Germline Mutations of TSH Receptor Gene as Cause of Nonautoimmune Subclinical Hypothyroidism. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 2549-2555.	1.8	127
10	Effect of gluten-free diet on bone mineral content in growing patients with celiac disease. American Journal of Clinical Nutrition, 1993, 57, 224-228.	2.2	124
11	Thyroid Nodules and Cancer in Children and Adolescents Affected by Autoimmune Thyroiditis. JAMA Pediatrics, 2008, 162, 526.	3.6	116
12	Congenital Hypothyroidism With Eutopic Thyroid Gland: Analysis of Clinical and Biochemical Features at Diagnosis and After Re-Evaluation. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 1395-1402.	1.8	107
13	A frequent oligogenic involvement in congenital hypothyroidism. Human Molecular Genetics, 2017, 26, 2507-2514.	1.4	107
14	Persistent mild hypothyroidism associated with novel sequence variants of theDUOX2 gene in two siblings. Human Mutation, 2005, 26, 395-395.	1.1	105
15	Bone Density and Bone Metabolism Are Normal After Long-Term Gluten-Free Diet in Young Celiac Patients. American Journal of Gastroenterology, 1999, 94, 398-403.	0.2	94
16	Bone density in young patients with congenital adrenal hyperplasia. Bone, 1996, 18, 337-340.	1.4	90
17	Genetics and phenomics of hypothyroidism due to TSH resistance. Molecular and Cellular Endocrinology, 2010, 322, 72-82.	1.6	87
18	Neonatal transient hypothyroidism: aetiological study. Archives of Disease in Childhood: Fetal and Neonatal Edition, 1998, 79, F70-F72.	1.4	80

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19	Diagnostic Features of Thyroid Nodules in Pediatrics. JAMA Pediatrics, 2010, 164, 714.	3.6	79
20	Evolution of Thyroid Function in Preterm Infants Detected by Screening for Congenital Hypothyroidism. Journal of Pediatrics, 2014, 164, 1296-1302.	0.9	78
21	Congenital Hypothyroidism due to Defects of Thyroid Development and Mild Increase of TSH at Screening: Data From the Italian National Registry of Infants With Congenital Hypothyroidism. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 1403-1408.	1.8	76
22	Molecular Analysis of the Pendredâ€™s Syndrome Gene and Magnetic Resonance Imaging Studies of the Inner Ear Are Essential for the Diagnosis of True Pendredâ€™s Syndrome1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 2469-2475.	1.8	75
23	The Clinical and Molecular Characterization of Patients With Dyshormonogenic Congenital Hypothyroidism Reveals Specific Diagnostic Clues for DUOX2 Defects. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E544-E553.	1.8	69
24	STUDY OF THE EFFECTS OF BROMOCRIPTINE ON SEXUAL IMPOTENCE. Clinical Endocrinology, 1977, 7, 417-421.	1.2	68
25	Sleep and upper airway obstruction in children with achondroplasia. Journal of Pediatrics, 1996, 129, 743-749.	0.9	66
26	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.	1.8	63
27	Mutational Analysis of GNAS1 in Patients with Pseudohypoparathyroidism: Identification of Two Novel Mutations1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 4243-4248.	1.8	57
28	Bone mass in young patients with type I diabetes. Bone and Mineral, 1990, 8, 23-30.	2.0	56
29	JAG1 Loss-Of-Function Variations as a Novel Predisposing Event in the Pathogenesis of Congenital Thyroid Defects. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 861-870.	1.8	54
30	Comparison of clinical-radiological and molecular findings in hypochondroplasia. , 1998, 75, 109-112.		52
31	Testicular microlithiasis: An unreported feature of McCUNE-Albright syndrome in males. Journal of Pediatrics, 2004, 145, 670-672.	0.9	49
32	Clinical heterogeneity and diagnostic delay of autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome. Clinical Immunology, 2011, 139, 6-11.	1.4	49
33	Longitudinal Changes of Bone Density and Bone Resorption in Hyperthyroid Girls During Treatment. Journal of Bone and Mineral Research, 1999, 14, 1971-1977.	3.1	48
34	Validation of Food Frequency Questionnaire for Assessing Dietary Macronutrients and Calcium Intake in Italian Children and Adolescents. Journal of Pediatric Gastroenterology and Nutrition, 2005, 40, 555-560.	0.9	47
35	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.	1.8	47
36	Congenital Hypothyroidism Treatment in Infants: A Comparative Study between Liquid and Tablet Formulations of Levothyroxine. Hormone Research in Paediatrics, 2014, 81, 50-54.	0.8	46

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37	Graves Disease in Children: Thyroid-Stimulating Hormone Receptor Antibodies as Remission Markers. Journal of Pediatrics, 2014, 164, 1189-1194.e1.	0.9	46
38	Neutralizing Anti-Cytokine Autoantibodies Against Interferon-Î± in Immunodysregulation Polyendocrinopathy Enteropathy X-Linked. Frontiers in Immunology, 2018, 9, 544.	2.2	46
39	Clinical and molecular heterogeneity in a large series of patients with hypophosphatemic rickets. Bone, 2015, 79, 143-149.	1.4	42
40	Newborn of mothers affected by autoimmune thyroiditis: the importance of thyroid function monitoring in the first months of life. Italian Journal of Pediatrics, 2010, 36, 24.	1.0	35
41	Serum Thyrotropin Concentration in Children with Isolated Thyroid Nodules. Journal of Pediatrics, 2013, 163, 1465-1470.	0.9	31
42	X-linked hypophosphatemic rickets: an Italian expertsâ€™ opinion survey. Italian Journal of Pediatrics, 2019, 45, 67.	1.0	31
43	Mild Hypothyroidism in Childhood: Who, When, and How Should Be Treated?. Journal of the Endocrine Society, 2018, 2, 1024-1039.	0.1	30
44	Effect of insulin treatment on osteocalcin levels in diabetic children and adolescents. Journal of Endocrinological Investigation, 1993, 16, 505-509.	1.8	29
45	Influence of gender and pubertal stage at diagnosis on growth outcome in childhood thyrotoxicosis: results of a collaborative study. Clinical Endocrinology, 2006, 64, 53-57.	1.2	29
46	The Italian screening program for primary congenital hypothyroidism: actions to improve screening, diagnosis, follow-up, and surveillance. Journal of Endocrinological Investigation, 2013, 36, 195-203.	1.8	29
47	Surgical management of pediatric Gravesâ€™ disease: an effective definitive treatment. Pediatric Surgery International, 2012, 28, 609-614.	0.6	28
48	Snyderâ€“Robinson syndrome: A novel nonsense mutation in spermine synthase and expansion of the phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2316-2320.	0.7	28
49	Autoimmune polyendocrine syndrome type 1: an Italian survey on 158 patients. Journal of Endocrinological Investigation, 2021, 44, 2493-2510.	1.8	28
50	Autosomal Dominant Hypocalcemia in Monozygotic Twins Caused by a De Novo Germline Mutation Near the Amino-Terminus of the Human Calcium Receptor. Journal of Bone and Mineral Research, 2004, 19, 578-586.	3.1	26
51	Clinical Presentation of McCune-Albright Syndrome in Males. Journal of Pediatric Endocrinology and Metabolism, 2006, 19, 619-22.	0.4	26
52	Genetic defects of hydrogen peroxide generation in the thyroid gland. Journal of Endocrinological Investigation, 2013, 36, 261-6.	1.8	26
53	Recombinant human TSH testing is a valuable tool for differential diagnosis of congenital hypothyroidism during l -thyroxine replacement. Clinical Endocrinology, 2003, 59, 230-236.	1.2	24
54	Asymptomatic Thyrotropin-Secreting Pituitary Macroadenoma in a 13-Year-Old Girl: Successful First-Line Treatment with Somatostatin Analogs. Thyroid, 2012, 22, 1076-1079.	2.4	24

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55	Neonatal Screening for Congenital Hypothyroidism: What Can We Learn From Discordant Twins?. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5765-5779.	1.8	24
56	Defective function of Fas in T cells from paediatric patients with autoimmune thyroid diseases. Clinical and Experimental Immunology, 2003, 133, 430-437.	1.1	23
57	Congenital hypothyroidism: Auxological retrospective study during the first six years of age. Journal of Endocrinological Investigation, 1996, 19, 224-229.	1.8	22
58	Thyroid scintigraphy and perchlorate test after recombinant human TSH: a new tool for the differential diagnosis of congenital hypothyroidism during infancy. European Journal of Nuclear Medicine and Molecular Imaging, 2007, 34, 1498-1503.	3.3	21
59	Neurophysiologic Studies and Cognitive Function in Congenital Hypothyroid Children. Pediatric Research, 1995, 37, 736-740.	1.1	20
60	Bone turnover in neonates: Changes of urinary excretion rate of collagen type I cross-linked peptides during the first days of life and influence of gestational age. Bone, 1997, 20, 563-566.	1.4	20
61	Diagnosis of hypochondroplasia: the role of radiological interpretation. Pediatric Radiology, 2001, 31, 203-208.	1.1	20
62	Total Iodide Organification Defect: Clinical and Molecular Characterization of an Italian Family. Thyroid, 2005, 15, 1085-1088.	2.4	20
63	Mild <scp>TSH</scp> resistance: Clinical and hormonal features in childhood and adulthood. Clinical Endocrinology, 2017, 87, 587-596.	1.2	20
64	Cognitive function and neurophysiological evaluation in early-treated hypothyroid children. Neurological Sciences, 2000, 21, 307-314.	0.9	19
65	Decreased Parietal Cortex Activity during Mental Rotation in Children with Congenital Hypothyroidism. Neuroendocrinology, 2009, 89, 56-65.	1.2	19
66	Bone Modeling Alteration in Premature Infants. JAMA Pediatrics, 1994, 148, 1215.	3.6	17
67	Levothyroxine Treatment in Pediatric Benign Thyroid Nodules. Hormone Research in Paediatrics, 2011, 75, 246-251.	0.8	17
68	Difficult treatment of consumptive hypothyroidism in a child with massive parotid hemangioma. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 153-5.	0.4	17
69	Genetic and epigenetic alterations in the GNAS locus and clinical consequences in Pseudohypoparathyroidism: Italian common healthcare pathways adoption. Italian Journal of Pediatrics, 2016, 42, 101.	1.0	17
70	Pediatric Pituitary Adenomas: Early and Long-Term Surgical Outcome in a Series of 85 Consecutive Patients. Neurosurgery, 2019, 85, 65-74.	0.6	17
71	Growth hormone therapy in children: predictive factors and short-term and long-term response criteria. Endocrine, 2019, 66, 614-621.	1.1	17
72	Human growth hormone treatment in prepubertal children with achondroplasia. , 1996, 61, 396-400.		16

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73	Brain Magnetic Resonance Imaging in Congenital Hypothyroid Infants at Diagnosis. Thyroid, 1997, 7, 761-764.	2.4	16
74	DUOXS defects: Genotype-phenotype correlations. Annales D'Endocrinologie, 2011, 72, 82-86.	0.6	16
75	Click-evoked otoacoustic emissions recorded from untreated congenital hypothyroid newborns. Hearing Research, 2002, 166, 136-142.	0.9	15
76	â€œBlock-and-replaceâ€•treatment in Gravesâ€™ disease: experience in a cohort of pediatric patients. Journal of Endocrinological Investigation, 2020, 43, 595-600.	1.8	14
77	Congenital hypothyroidism with gland in situ: Diagnostic re-evaluation. Journal of Endocrinological Investigation, 2005, 28, 516-522.	1.8	13
78	Pseudohypoparathyroidism, an often delayed diagnosis: a case series. Cases Journal, 2009, 2, 6734.	0.4	13
79	Suprasellar granular cell tumor of the neurohypophysis in a child: unusual presentation in pediatric age of a rare tumor. Child's Nervous System, 2013, 29, 1031-1034.	0.6	13
80	NovelNKX2-1Frameshift Mutations in Patients with Atypical Phenotypes of the Brain-Lung-Thyroid Syndrome. European Thyroid Journal, 2014, 3, 227-33.	1.2	13
81	Final height in Italian patients with congenital hypothyroidism detected by neonatal screening: a 20-year observational study. Italian Journal of Pediatrics, 2015, 41, 82.	1.0	13
82	Levothyroxine requirement in congenital hypothyroidism: a 12-year longitudinal study. Endocrine, 2015, 50, 674-680.	1.1	12
83	Thyroid function in children and adolescents with Hashimoto's thyroiditis after l-thyroxine discontinuation. Endocrine Connections, 2017, 6, 206-212.	0.8	12
84	Heterogeneous phenotype in children affected by non-autoimmune hypothyroidism: an update. Journal of Endocrinological Investigation, 2015, 38, 835-840.	1.8	11
85	Newborn Screening for Congenital Hypothyroidism: the Benefit of Using Differential TSH Cutoffs in a 2-Screen Program. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e338-e349.	1.8	11
86	Vitamin D dependent rickets, diagnostic and therapeutic difficulties: two case reports. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 801-5.	0.4	6
87	Prevalence of activating thyrotropin receptor and Gsî± gene mutations in paediatric thyroid toxic adenomas: a multicentric Italian study. Clinical Endocrinology, 2013, 79, 747-749.	1.2	5
88	Early Fractures and Occult Hyperthyroidism: McCune-Albright Syndrome?. Hormone Research in Paediatrics, 2001, 56, 58-62.	0.8	4
89	Severe hypocalcemia due to a de novo mutation in the fifth transmembrane domain of the calcium-sensing receptor. American Journal of Medical Genetics, Part A, 2006, 140A, 98-101.	0.7	4
90	Absence of sonic hedgehog (Shh) germline mutations in patients with thyroid dysgenesis. Clinical Endocrinology, 2008, 69, 828-829.	1.2	4

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91	Treatment of Congenital Hypothyroidism: Comparison Between L-Thyroxine Oral Solution and Tablet Formulations up to 3 years of age. European Journal of Endocrinology, 2021, 186, 45-52.	1.9	4
92	Rare cases of autoimmune hypothyroidism in young children. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 963-6.	0.4	2
93	Pseudohypoparathyroidism: application of the Italian common healthcare-pathway for a homogeneous clinical approach and a shared follow up. Italian Journal of Pediatrics, 2021, 47, 48.	1.0	2
94	Linear growth and puberty in childhood obesity: what is new?. Minerva Pediatrics, 2022, 73, .	0.2	2
95	Type 1 Polyglandular Autoimmune Syndrome in a Girl: Diagnostic and Therapeutic Problems. Journal of Pediatric Endocrinology and Metabolism, 1989, 3, .	0.4	0
96	Transient Neonatal Hypothyroidism. , 2015, , 75-83.		0
97	Endocrinology: Diagnostics in Children and Adolescents. , 2016, , 127-138.		0