Marek Niedziela

List of Publications by Year
in descending order

95 papers	2,260 citations	304368 22 h-index	233125 45 g-index
100 all docs	100 docs citations	100 times ranked	3084 citing authors

#	Article	IF	Citations
1	<emph type="ital">DICER1</emph> Mutations in Familial Multinodular Goiter With and Without Ovarian Sertoli-Leydig Cell Tumors. JAMA - Journal of the American Medical Association, 2011, 305, 68.	3.8	284
2	Pathogenesis, diagnosis and management of thyroid nodules in children. Endocrine-Related Cancer, 2006, 13, 427-453.	1.6	270
3	Mutations in FGF17, IL17RD, DUSP6, SPRY4, and FLRT3 Are Identified in Individuals with Congenital Hypogonadotropic Hypogonadism. American Journal of Human Genetics, 2013, 92, 725-743.	2.6	227
4	Changes Over Time in Sex Assignment for Disorders of Sex Development. Pediatrics, 2014, 134, e710-e715.	1.0	98
5	Clinical Relevance of Thyroid-Stimulating Autoantibodies in Pediatric Graves' Diseaseâ€"A Multicenter Study. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 1648-1655.	1.8	92
6	Rola ukÅ‚adu immunologicznego oraz udziaÅ‚ cytokin w patomechanizmie autoimmunologicznej choroby tarczycy (AITD). Endokrynologia Polska, 2014, 65, 150-155.	0.3	92
7	Galectin-3 Is Not an Universal Marker of Malignancy in Thyroid Nodular Disease in Children and Adolescents. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4411-4415.	1.8	67
8	Hot Nodules in Children and Adolescents in Western Poland from 1996 to 2000: Clinical Analysis of 31 Patients. Journal of Pediatric Endocrinology and Metabolism, 2002, 15, 823-30.	0.4	66
9	Recurrent EZH1 mutations are a second hit in autonomous thyroid adenomas. Journal of Clinical Investigation, 2016, 126, 3383-3388.	3.9	66
10	Increased risk of thyroid pathology in patients with thyroid hemiagenesis: results of a large cohort caseâ€"control study. European Journal of Endocrinology, 2010, 162, 153-160.	1.9	57
11	Thyroid nodules. Best Practice and Research in Clinical Endocrinology and Metabolism, 2014, 28, 245-277.	2.2	47
12	A prospective study of thyroid nodular disease in children and adolescents in western Poland from 1996 to 2000 and the incidence of thyroid carcinoma relative to iodine deficiency and the Chernobyl disaster. Pediatric Blood and Cancer, 2004, 42, 84-92.	0.8	42
13	2012 European Thyroid Association Guidelines for the Management of Familial and Persistent Sporadic Non-Autoimmune Hyperthyroidism Caused by Thyroid-Stimulating Hormone Receptor Germline Mutations. European Thyroid Journal, 2012, 1, 142-147.	1.2	38
14	Significant Benefits of AIP Testing and Clinical Screening in Familial Isolated and Young-onset Pituitary Tumors. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2247-e2260.	1.8	37
15	Germ-line deletion in DICER1 revealed by a novel MLPA assay using synthetic oligonucleotides. European Journal of Human Genetics, 2014, 22, 564-567.	1.4	35
16	Somatic mutations in 33 benign and malignant hot thyroid nodules in children and adolescents. Molecular and Cellular Endocrinology, 2014, 393, 39-45.	1.6	32
17	Rekomendacje Polskich Towarzystw Naukowych â€žDiagnostyka i leczenie raka tarczycyâ€• Aktualizacja na rok 2018. Endokrynologia Polska, 2018, 69, 34-74.	0.3	32
18	Diagnostyka i leczenie raka tarczycy. Endokrynologia Polska, 2016, 67, 74-145.	0.3	31

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19	FOXE1 Polyalanine Tract Length Polymorphism in Patients with Thyroid Hemiagenesis and Subjects with Normal Thyroid. Hormone Research in Paediatrics, 2011, 75, 329-334.	0.8	30
20	The clinical diagnosis and molecular genetics of kearns-sayre syndrome: a complex mitochondrial encephalomyopathy. Pediatric Endocrinology Reviews, 2006, 4, 117-37.	1.2	28
21	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
22	The clinical role of serum concentrations of selected cytokines: IL-1Î², TNF-Î±and IL-6 in diagnosis of autoimmune thyroid disease (AITD) in children. Autoimmunity, 2014, 47, 466-472.	1.2	24
23	Adult-Onset Cervical Embryonal Rhabdomyosarcoma and DICER1 Mutations. Journal of Lower Genital Tract Disease, 2016, 20, e8-e10.	0.9	23
24	Clinical and Molecular Characteristics May Alter Treatment Strategies of Thyroid Malignancies in DICER1 Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 277-284.	1.8	22
25	Polskie rekomendacje diagnostyki i leczenia zrÃ³Å¼nicowanego raka tarczycy u dzieci. Endokrynologia Polska, 2016, 67, 628-642.	0.3	22
26	No Correlation between Androgen Receptor CAG and GGN Repeat Length and the Degree of Genital Virilization in Females with 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 2443-2450.	1.8	21
27	A novel mitochondrial DNA deletion in a patient with Kearns-Sayre syndrome: a late-onset of the fatal cardiac conduction deficit and cardiomyopathy accompanying long-term rGH treatment. BMC Pediatrics, 2013, 13, 27.	0.7	19
28	Evidence of a significant vitamin D deficiency among 9â€“13-year-old Polish children: results of a multicentre study. European Journal of Nutrition, 2019, 58, 2029-2036.	1.8	19
29	Severe Hypothyroidism Due to Autoimmune Atrophic Thyroiditis - Predicted Target Height and a Plausible Mechanism for Sexual Precocity. Journal of Pediatric Endocrinology and Metabolism, 2001, 14, 901-7.	0.4	18
30	Exome sequencing reveals two novel compound heterozygous XYLT1 mutations in a Polish patient with Desbuquois dysplasia type 2 and growth hormone deficiency. Journal of Human Genetics, 2016, 61, 577-583.	1.1	18
31	Mutations in proteasome-related genes are associated with thyroid hemiagenesis. Endocrine, 2017, 56, 279-285.	1.1	18
32	Integrating clinical and genetic approaches in the diagnosis of 46,XY disorders of sex development. Endocrine Connections, 2018, 7, 1480-1490.	0.8	18
33	Direct effects of the pineal hormone melatonin on testosterone synthesis of Leydig cells in Djungarian hamsters (Phodopus sungorus) in vitro. Neuroscience Letters, 1995, 201, 247-250.	1.0	17
34	Birth Weight in Different Etiologies of Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1044-1050.	1.8	16
35	Usefulness of Growth Hormone (GH) Stimulation Tests and IGF-1 Concentration Measurement in GH Deficiency Diagnosis. Journal of Pediatric Endocrinology and Metabolism, 2008, 21, 569-580.	0.4	16
36	Virilizing ovarian tumor in a 14â€yearâ€old female with a prior familial multinodular goiter. Pediatric Blood and Cancer, 2008, 51, 543-545.	0.8	15

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37	Proinflammatory Cytokines in Monitoring the Course of Disease and Effectiveness of Treatment with Etanercept (ETN) of children with Oligo- and Polyarticular Juvenile Idiopathic Arthritis (JIA). Clinical Laboratory, 2014, 60, 1481-90.	0.2	15
38	Thyroid carcinoma in a fourteen-year-old boy with Graves disease. Medical and Pediatric Oncology, 2002, 38, 290-291.	1.0	13
39	Authorsâ€™ Response: Methodological Considerations Regarding the Use of Galectin-3 Expression Analysis in Preoperative Evaluation of Thyroid Nodules. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 950-951.	1.8	13
40	NR0B1 (DAX1) mutations in patients affected by congenital adrenal hypoplasia with growth hormone deficiency as a new finding. Journal of Applied Genetics, 2013, 54, 225-230.	1.0	13
41	SEMA3A and IGSF10 Are Novel Contributors to Combined Pituitary Hormone Deficiency (CPHD). Frontiers in Endocrinology, 2020, 11, 368.	1.5	13
42	Expression of Bcl-2 Family Proteins in Thyrocytes from Young Patients with Immune and Nonimmune Thyroid Diseases. Hormone Research, 2008, 70, 155-164.	1.8	12
43	Molecular Detection and Incidence of Y Chromosomal Material in Patients with Turner Syndrome. Sexual Development, 2017, 11, 254-261.	1.1	10
44	The Usefulness of Magnetic Resonance Imaging of the Cardiovascular System in the Diagnostic Work-Up of Patients With Turner Syndrome. Frontiers in Endocrinology, 2018, 9, 609.	1.5	10
45	Gonadectomy in conditions affecting sex development: a registry-based cohort study. European Journal of Endocrinology, 2021, 184, 791-801.	1.9	9
46	X-Linked Adrenal Hypoplasia Congenita in a Boy due to a Novel Deletion of the EntireNR0B1 (DAX1)andMAGEB1â€“4Genes. International Journal of Endocrinology, 2016, 2016, 1-7.	0.6	8
47	Familial central precocious puberty: two novel MKRN3 mutations. Pediatric Research, 2020, 90, 431-435.	1.1	8
48	Mosaic IL6ST variant inducing constitutive GP130 cytokine receptor signaling as a cause of neonatal onset immunodeficiency with autoinflammation and dysmorphy. Human Molecular Genetics, 2021, 30, 226-233.	1.4	8
49	Papillary Thyroid Carcinoma in a Boy with Familial Tuberous Sclerosis Complex Attributable to a TSC2 Deletionâ€”A Case Report. Current Oncology, 2017, 24, 423-428.	0.9	7
50	Limited Mandibular Movements as a Consequence of Unilateral or Asymmetrical Temporomandibular Joint Involvement in Juvenile Idiopathic Arthritis Patients. Journal of Clinical Medicine, 2020, 9, 2576.	1.0	7
51	Adrenal Function in Children with Severe Asthma Treated with High-Dose Inhaled Glucocorticoids: Recommended Screening Tests in Outpatient Conditions. Journal of Pediatric Endocrinology and Metabolism, 2007, 20, 781-9.	0.4	6
52	Subclinical hypothyroidism: Dilemmas in the treatment of children. Journal of Endocrinological Investigation, 2007, 30, 529-531.	1.8	6
53	Uterine Development During Induced Puberty in Girls with Turner Syndrome. Frontiers in Endocrinology, 2021, 12, 707031.	1.5	6
54	Mental health and adjustment to juvenile idiopathic arthritis: Level of agreement between parent and adolescent reports according to Strengths and Difficulties Questionnaire and Adolescent Outcomes Questionnaire. PLoS ONE, 2017, 12, e0173768.	1.1	6

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55	PAPP-A2 nowy regulator procesu wzrastania. Endokrynologia Polska, 2017, 68, 682-691.	0.3	6
56	Serum TNF-Î± levels and Indicators of Disease Activity in Children with Oligoarticular Juvenile Idiopathic Arthritis (oJIA) in the First Year of the Disease. Clinical Laboratory, 2014, 60, 799-807.	0.2	6
57	Growth, puberty and testicular function in boys born small for gestational age with a nonspecific disorder of sex development. Clinical Endocrinology, 2022, 96, 165-174.	1.2	6
58	A novel mutation in the NR0B1 (DAX1) gene in a large family with two boys affected by congenital adrenal hypoplasia. Hormones, 2002, 13, 413-9.	0.9	5
59	Autoimmune thyroid disease and allergic contact dermatitis: two immune-related pathologies in the same patient. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 31-2.	0.4	5
60	Treatment of cartilageâ€“hair hypoplasia with recombinant human growth hormone. Pediatrics International, 2013, 55, e162-4.	0.2	5
61	The influence of growth hormone therapy on the cardiovascular system in Turner syndrome. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1363-1372.	0.4	5
62	Pituitary Microsomal Autoantibodies in Patients with Childhood-Onset Combined Pituitary Hormone Deficiency: an Antigen Identification Attempt. Archivum Immunologiae Et Therapiae Experimentalis, 2016, 64, 485-495.	1.0	4
63	Diagnostic significance of serum concentrations of soluble Fas ligand (sFasL) in children with autoimmune thyroid disease. Autoimmunity, 2017, 50, 192-198.	1.2	4
64	The Pediatric Outcomes Data Collection Instrument for a Polish sample with juvenile idiopathic arthritis: psychometric properties of proxy version. International Journal of Rheumatic Diseases, 2017, 20, 2077-2085.	0.9	4
65	Multiples of Median-Transformed, Normalized Reference Ranges of Steroid Profiling Data Independent of Age, Sex, and Units. Hormone Research in Paediatrics, 2018, 89, 255-264.	0.8	4
66	Testosterone Therapy and Its Monitoring in Adolescent Boys with Hypogonadism: Results of an International Survey from the I-DSD Registry. Sexual Development, 2021, 15, 236-243.	1.1	4
67	Hyperthyroidism in adolescents. Endocrine Connections, 2021, 10, R279-R292.	0.8	4
68	Letter to the Editor Amiodarone-induced thyrotoxicosis with paroxysmal supraventricular (ectopic) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.4	3
69	AlstrÃ¶m syndrome: A case report of the Polish family and a brief review of the differential diagnosis. Pediatria Polska, 2017, 92, 781-785.	0.1	3
70	CDON gene contributes to pituitary stalk interruption syndrome associated with unilateral facial and abducens nerve palsy. Journal of Applied Genetics, 2021, 62, 621-629.	1.0	3
71	Growth hormone therapy in a girl with Turner syndrome and diabetes type 1 â€“ case report. Pediatric Endocrinology, Diabetes and Metabolism, 2014, 20, 75-81.	0.3	3
72	Syndrome of Congenital Insulin Resistance Caused by a Novel INSR Gene Mutation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2023, 15, 312-317.	0.4	3

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73	Isolation method of Leydig cells from mature male Djungarian hamsters (Phodopus sungorus) and their steroidogenic activity in vitro. Andrologia, 1999, 31, 157-161.	1.0	2
74	Insulin Receptor and its Relationship with Different Forms of Insulin Resistance. Advances in Cell Biology, 2010, 2, 59-90.	1.5	2
75	Growth Impairment in a Boy with Late-Onset Congenital Adrenal Hyperplasia and Anorexia Nervosa. Experimental and Clinical Endocrinology and Diabetes, 2010, 118, 180-183.	0.6	2
76	Diabetes insipidus coexisting with sellar-suprasellar tumor â€“. Wspolczesna Onkologia, 2011, 6, 403-406.	0.7	2
77	X-linked hypophosphataemic rickets in children: clinical phenotype, therapeutic strategies, and molecular background. Endokrynologia Polska, 2021, 72, 108-119.	0.3	2
78	Letter to the Editor Adrenal function and MC1R gene analysis in a prepubertal girl with generalized hyperpigmentation:. Archives of Medical Science, 2013, 4, 761-764.	0.4	1
79	Kawasaki-like syndrome in children from Greater Poland during the first wave of COVID-19 pandemic. Pediatria I Medycyna Rodzinna, 2020, 16, 396-403.	2.3	1
80	Is growth without IGF1 possible? A case report. Pediatric Endocrinology, Diabetes and Metabolism, 2017, 23, 215-220.	0.3	1
81	PrzydatnoÅ›Ä‡ testu GHRH w diagnostyce niedoboru hormonu wzrostu u dzieci. Endokrynologia Polska, 2015, 66, 137-141.	0.3	1
82	Newborns of mothers with autoimmune thyroid disease: Safe or at risk?. Early Human Development, 2008, 84, S127.	0.8	0
83	Bridging clinical care and basic research. Pediatric Health, 2010, 4, 571-577.	0.3	0
84	The usefulness of ultrasound in follow-up of a patient with dyshormonogenetic congenital hypothyroidism. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 549-50.	0.4	0
85	Somatic mutations in 29 hot nodules in children. Experimental and Clinical Endocrinology and Diabetes, 2013, 121, .	0.6	0
86	Genome-wide survey for clinically relevant structural abnormalities contributing to pathogenesis of combined pituitary hormone deficiency (CPHD) with childhood onset.. Endocrine Abstracts, 0, , .	0.0	0
87	The increased coexistence of autoimmune thyroiditis (AIT) in children and adolescents with thyroid carcinoma (TC) in years 2001-2015 compared to years 1996-2000. Endocrine Abstracts, 0, , .	0.0	0
88	Analysis of a large cohort of subjects with thyroid hemiagenesis (THA) reveals random seasonality in the dates of birth. Endocrine Abstracts, 0, , .	0.0	0
89	Analysis of the Seasonality of Births in a Large Cohort of Patients with Thyroid Hemiagenesis - A Preliminary Study. Iranian Journal of Pediatrics, 2018, 28, .	0.1	0
90	Gonadectomy for adults with DSD conditions at risk of hypogonadism in the international disorders of sex development registry. Endocrine Abstracts, 0, , .	0.0	0

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91	Hyperthyroidism in adolescents. Endocrine Abstracts, 0, , .	0.0	0
92	Intrathyroidal Thymus (Incidentaloma) Mimicking Thyroid Neoplasia in DICER1 Syndrome. European Thyroid Journal, 2021, 10, 257-261.	1.2	0
93	Intrathyroidal Thymus (Incidentaloma) Mimicking Thyroid Neoplasia in DICER1 Syndrome. European Thyroid Journal, 2021, 10, 257-261.	1.2	0
94	Moderate congenital adrenal hyperplasia in two girls diagnosed by newborn screening. Pediatric Endocrinology, Diabetes and Metabolism, 2021, 27, 291-297.	0.3	0
95	NKX2-5 Variant in Two Siblings with Thyroid Hemiagenesis. International Journal of Molecular Sciences, 2022, 23, 3414.	1.8	0