Nelly Pitteloud

List of Publications by Year
in descending order

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119 papers	10,230 citations	36691 53 h-index	39744 98 g-index
124 all docs	124 docs citations	124 times ranked	6336 citing authors

#	Article	IF	Citations
1	Longitudinal evaluation of multiple biomarkers for the detection of testosterone gel administration in women with normal menstrual cycle. Drug Testing and Analysis, 2022, 14, 833-850.	1.6	29
2	Long-term body composition improvement in post-menopausal women following bariatric surgery: a cross-sectional and caseâ€“control study. European Journal of Endocrinology, 2022, 186, 255-263.	1.9	2
3	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	1.1	14
4	Transcriptome profiling of kisspeptin neurons from the mouse arcuate nucleus reveals new mechanisms in estrogenic control of fertility. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	11
5	Kisspeptin-54 Accurately Identifies Hypothalamic Gonadotropin-Releasing Hormone Neuronal Dysfunction in Men with Congenital Hypogonadotropic Hypogonadism. Neuroendocrinology, 2021, 111, 1176-1186.	1.2	12
6	Congenital Hypogonadotropic Hypogonadism with Anosmia and Gorlin Features Caused by a PTCH1 Mutation Reveals a New Candidate Gene for Kallmann Syndrome. Neuroendocrinology, 2021, 111, 99-114.	1.2	20
7	Compromised Volumetric Bone Density and Microarchitecture in Men With Congenital Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e3312-e3326.	1.8	10
8	Testosterone-induced increase in libido in a patient with a loss-of-function mutation in the AR gene. Endocrinology, Diabetes and Metabolism Case Reports, 2021, 2021, .	0.2	0
9	Steroid profiling by UHPLC-MS/MS in dried blood spots collected from healthy women with and without testosterone gel administration. Journal of Pharmaceutical and Biomedical Analysis, 2021, 204, 114280.	1.4	24
10	Hypothalamic Disorders During Ovulation, Pregnancy, and Lactation. , 2020, , 217-240.		0
11	Neuron-Derived Neurotrophic Factor Is Mutated in Congenital Hypogonadotropic Hypogonadism. American Journal of Human Genetics, 2020, 106, 58-70.	2.6	39
12	Pathogenic mosaic variants in congenital hypogonadotropic hypogonadism. Genetics in Medicine, 2020, 22, 1759-1767.	1.1	7
13	Precision medicine in the era of artificial intelligence: implications in chronic disease management. Journal of Translational Medicine, 2020, 18, 472.	1.8	99
14	Non-classic cytochrome P450 oxidoreductase deficiency strongly linked with menstrual cycle disorders and female infertility as primary manifestations. Human Reproduction, 2020, 35, 939-949.	0.4	13
15	Non-invasive assessment of coronary endothelial function in children and adolescents with type 1 diabetes mellitus using isometric handgrip exerciseâ€"MRI: A feasibility study. PLoS ONE, 2020, 15, e0228569.	1.1	5
16	Congenital hypogonadotropic hypogonadism/Kallmann syndrome is associated with statural gain in both men and women: a monocentric study. European Journal of Endocrinology, 2020, 182, 185.	1.9	21
17	SAT-010 Non-Classic POR Deficiency as a Cause of Menstrual Disorders & Infertility. Journal of the Endocrine Society, 2020, 4, .	0.1	0
18	Functional Hypogonadotropic Hypogonadism in Men: Underlying Neuroendocrine Mechanisms and Natural History. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3403-3414.	1.8	28

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19	Clomiphene citrate effect on testosterone level and semen parameters in 18 infertile men with low testosterone level and normal/low gonadotropines level. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 238, 104-109.	0.5	14
20	Congenital Hypogonadotropic Hypogonadism (Isolated GnRH Deficiency). Contemporary Endocrinology, 2019, , 229-250.	0.3	3
21	Clinical Management of Congenital Hypogonadotropic Hypogonadism. Endocrine Reviews, 2019, 40, 669-710.	8.9	244
22	Combined immune checkpoint inhibitor therapy with nivolumab and ipilimumab causing acute-onset type 1 diabetes mellitus following a single administration: two case reports. BMC Endocrine Disorders, 2019, 19, 144.	0.9	24
23	Thyrotropin-secreting pituitary adenomas: a systematic review and meta-analysis of postoperative outcomes and management. Pituitary, 2019, 22, 79-88.	1.6	36
24	A novel CHD7 mutation in an adolescent presenting with growth and pubertal delay. Annals of Pediatric Endocrinology and Metabolism, 2019, 24, 49-54.	0.8	5
25	Congenital hypogonadotropic hypogonadism and constitutional delay of growth and puberty have distinct genetic architectures. European Journal of Endocrinology, 2018, 178, 377-388.	1.9	95
26	DCC/NTN1 complex mutations in patients with congenital hypogonadotropic hypogonadism impair GnRH neuron development. Human Molecular Genetics, 2018, 27, 359-372.	1.4	42
27	Evaluating CHARGE syndrome in congenital hypogonadotropic hypogonadism patients harboring CHD7 variants. Genetics in Medicine, 2018, 20, 872-881.	1.1	38
28	Intraoperative 2D C-arm and 3D O-arm in children: A comparative phantom study. Journal of Children's Orthopaedics, 2018, 12, 550-557.	0.4	13
29	Î²-Klotho deficiency shifts the gut-liver bile acid axis and induces hepatic alterations in mice. American Journal of Physiology - Endocrinology and Metabolism, 2018, 315, E833-E847.	1.8	13
30	Accuracy, satisfaction and usability of a flash glucose monitoring system among children and adolescents with type 1 diabetes attending a summer camp. Pediatric Diabetes, 2018, 19, 1276-1284.	1.2	20
31	The Reduction of Visceral Adipose Tissue after Roux-en-Y Gastric Bypass Is more Pronounced in Patients with Impaired Glucose Metabolism. Obesity Surgery, 2018, 28, 4006-4013.	1.1	16
32	Transition of Care from Childhood to Adulthood: Congenital Hypogonadotropic Hypogonadism. Endocrine Development, 2018, 33, 82-98.	1.3	6
33	Integrating clinical and genetic approaches in the diagnosis of 46,XY disorders of sex development. Endocrine Connections, 2018, 7, 1480-1490.	0.8	18
34	REV-ERBÎ± regulates Fgf21 expression in the liver via hepatic nuclear factor 6. Biology Open, 2017, 6, 1-7.	0.6	14
35	Genetic testing facilitates prepubertal diagnosis of congenital hypogonadotropic hypogonadism. Clinical Genetics, 2017, 92, 213-216.	1.0	14
36	Defective signaling through plexin-A1 compromises the development of the peripheral olfactory system and neuroendocrine reproductive axis in mice. Human Molecular Genetics, 2017, 26, 2006-2017.	1.4	44

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37	The induction of ovulation by pulsatile administration of GnRH: an appropriate method in hypothalamic amenorrhea. Gynecological Endocrinology, 2017, 33, 598-601.	0.7	18
38	<scp>KLB</scp> , encoding Î²â€Klotho, is mutated in patients with congenital hypogonadotropic hypogonadism. EMBO Molecular Medicine, 2017, 9, 1379-1397.	3.3	77
39	Two missense mutations in KCNQ1 cause pituitary hormone deficiency and maternally inherited gingival fibromatosis. Nature Communications, 2017, 8, 1289.	5.8	33
40	Beyond hormone replacement: quality of life in women with congenital hypogonadotropic hypogonadism. Endocrine Connections, 2017, 6, 404-412.	0.8	31
41	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
42	Adherence to treatment in men with hypogonadotrophic hypogonadism. Clinical Endocrinology, 2017, 86, 377-383.	1.2	32
43	Î²-Klotho deficiency protects against obesity through a crosstalk between liver, microbiota, and brown adipose tissue. JCI Insight, 2017, 2, .	2.3	41
44	Glycaemic, blood pressure and lipid goal attainment and chronic kidney disease stage of type 2 diabetic patients treated in primary care practices. Swiss Medical Weekly, 2017, 147, w14459.	0.8	7
45	Surgical treatment of acromegaly according to the 2010 remission criteria: systematic review and meta-analysis. Acta Neurochirurgica, 2016, 158, 2109-2121.	0.9	42
46	MANAGEMENT OF ENDOCRINE DISEASE: Reversible hypogonadotropic hypogonadism. European Journal of Endocrinology, 2016, 174, R267-R274.	1.9	66
47	Chronic kidney disease in type 2 diabetic patients followed-up by primary care physicians in Switzerland: prevalence and prescription of antidiabetic drugs. Swiss Medical Weekly, 2016, 146, w14282.	0.8	8
48	Psychosexual Development in Men with Congenital Hypogonadotropic Hypogonadism on Long-Term Treatment: A Mixed Methods Study. Sexual Medicine, 2015, 3, 32-41.	0.9	34
49	TRANSITION IN ENDOCRINOLOGY: Hypogonadism in adolescence. European Journal of Endocrinology, 2015, 173, R15-R24.	1.9	59
50	European Consensus Statement on congenital hypogonadotropic hypogonadismâ€"pathogenesis, diagnosis and treatment. Nature Reviews Endocrinology, 2015, 11, 547-564.	4.3	664
51	FGFR1 and PROKR2 rare variants found in patients with combined pituitary hormone deficiencies. Endocrine Connections, 2015, 4, 100-107.	0.8	34
52	Gonadotrophin replacement for induction of fertility in hypogonadal men. Best Practice and Research in Clinical Endocrinology and Metabolism, 2015, 29, 91-103.	2.2	96
53	Congenital hypogonadotropic hypogonadism with split hand/foot malformation: a clinical entity with a high frequency of FGFR1 mutations. Genetics in Medicine, 2015, 17, 651-659.	1.1	55
54	Mid-gut ACTH-secreting neuroendocrine tumor unmasked with 18F-dihydroxyphenylalanine-positron emission tomography. Endocrinology, Diabetes and Metabolism Case Reports, 2015, 2015, 140104.	0.2	2

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55	Reversal and Relapse of Hypogonadotropic Hypogonadism: Resilience and Fragility of the Reproductive Neuroendocrine System. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 861-870.	1.8	144
56	Hormonal control of spermatogenesis in men: Therapeutic aspects in hypogonadotropic hypogonadism. Annales D'Endocrinologie, 2014, 75, 98-100.	0.6	21
57	Mutation screening of SEMA3A and SEMA7A in patients with congenital hypogonadotropic hypogonadism. Pediatric Research, 2014, 75, 641-644.	1.1	64
58	Identifying the unmet health needs of patients with congenital hypogonadotropic hypogonadism using a web-based needs assessment: implications for online interventions and peer-to-peer support. Orphanet Journal of Rare Diseases, 2014, 9, 83.	1.2	63
59	Comparative functional analysis of two fibroblast growth factor receptor 1 (FGFR1) mutations affecting the same residue (R254W and R254Q) in isolated hypogonadotropic hypogonadism (IHH). Gene, 2013, 516, 146-151.	1.0	19
60	Abrupt decrease in serum testosterone levels after an oral glucose load in men: implications for screening for hypogonadism. Clinical Endocrinology, 2013, 78, 291-296.	1.2	91
61	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
62	Trial of Recombinant Follicle-Stimulating Hormone Pretreatment for GnRH-Induced Fertility in Patients with Congenital Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1790-E1795.	1.8	124
63	Responsiveness to a Physiological Regimen of GnRH Therapy and Relation to Genotype in Women With Isolated Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E206-E216.	1.8	24
64	Prioritizing Genetic Testing in Patients With Kallmann Syndrome Using Clinical Phenotypes. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E943-E953.	1.8	157
65	Variations inPROKR2, But NotPROK2, Are Associated With Hypopituitarism and Septo-optic Dysplasia. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E547-E557.	1.8	54
66	Testosterone restoration using enclomiphene citrate in men with secondary hypogonadism: a pharmacodynamic and pharmacokinetic study. BJU International, 2013, 112, 1188-1200.	1.3	35
67	When Genetic Load Does Not Correlate with Phenotypic Spectrum: Lessons from the GnRH Receptor (GNRHR). Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1798-E1807.	1.8	43
68	Olfactory Phenotypic Spectrum in Idiopathic Hypogonadotropic Hypogonadism: Pathophysiological and Genetic Implications. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E136-E144.	1.8	100
69	Klotho Coreceptors Inhibit Signaling by Paracrine Fibroblast Growth Factor 8 Subfamily Ligands. Molecular and Cellular Biology, 2012, 32, 1944-1954.	1.1	74
70	Genetic Overlap in Kallmann Syndrome, Combined Pituitary Hormone Deficiency, and Septo-Optic Dysplasia. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E694-E699.	1.8	136
71	An ancient founder mutation in PROKR2 impairs human reproduction. Human Molecular Genetics, 2012, 21, 4314-4324.	1.4	31
72	A Genetic Basis for Functional Hypothalamic Amenorrhea. New England Journal of Medicine, 2011, 364, 215-225.	13.9	219

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73	Estradiol levels in men with congenital hypogonadotropic hypogonadism and the effects ofÂdifferent modalities of hormonal treatment. Fertility and Sterility, 2011, 95, 2324-2329.e3.	0.5	30
74	The puzzles of the prokineticin 2 pathway in human reproduction. Molecular and Cellular Endocrinology, 2011, 346, 44-50.	1.6	19
75	Role of fibroblast growth factor (FGF) signaling in the neuroendocrine control of human reproduction. Molecular and Cellular Endocrinology, 2011, 346, 37-43.	1.6	24
76	Genetic basis and variable phenotypic expression of Kallmann syndrome: towards a unifying theory. Trends in Endocrinology and Metabolism, 2011, 22, 249-58.	3.1	127
77	The Role of the Prokineticin 2 Pathway in Human Reproduction: Evidence from the Study of Human and Murine Gene Mutations. Endocrine Reviews, 2011, 32, 225-246.	8.9	95
78	GnRH-Deficient Phenotypes in Humans and Mice with Heterozygous Variants inKISS1/Kiss1. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1771-E1781.	1.8	59
79	Heparan sulfate 6-O-sulfotransferase 1 , a gene involved in extracellular sugar modifications, is mutated in patients with idiopathic hypogonadotrophic hypogonadism. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11524-11529.	3.3	153
80	NovelFGF8Mutations Associated with Recessive Holoprosencephaly, Craniofacial Defects, and Hypothalamo-Pituitary Dysfunction. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1709-E1718.	1.8	113
81	Expanding the Phenotype and Genotype of Female GnRH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E566-E576.	1.8	97
82	Congenital Idiopathic Hypogonadotropic Hypogonadism: Evidence of Defects in the Hypothalamus, Pituitary, and Testes. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3019-3027.	1.8	115
83	Deciphering Genetic Disease in the Genomic Era: The Model of GnRH Deficiency. Science Translational Medicine, 2010, 2, 32rv2.	5.8	48
84	Oligogenic basis of isolated gonadotropin-releasing hormone deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 15140-15144.	3.3	313
85	Human GnRH Deficiency: A Unique Disease Model to Unravel the Ontogeny of GnRH Neurons. Neuroendocrinology, 2010, 92, 81-99.	1.2	87
86	Complex Genetics in Idiopathic Hypogonadotropic Hypogonadism. Frontiers of Hormone Research, 2010, 39, 142-153.	1.0	57
87	Nonsense Mutations inFGF8Gene Causing Different Degrees of Human Gonadotropin-Releasing Deficiency. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3491-3496.	1.8	70
88	The Long-Term Clinical Follow-Up and Natural History of Men with Adult-Onset Idiopathic Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 4235-4243.	1.8	45
89	Role of Seminiferous Tubular Development in Determining the FSH versus LH Responsiveness to GnRH in Early Sexual Maturation. Neuroendocrinology, 2009, 90, 260-268.	1.2	9
90	GNRH1 mutations in patients with idiopathic hypogonadotropic hypogonadism. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 11703-11708.	3.3	169

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91	Impaired Fibroblast Growth Factor Receptor 1 Signaling as a Cause of Normosmic Idiopathic Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4380-4390.	1.8	82
92	CHD7 mutations in patients initially diagnosed with Kallmann syndrome â€“ the clinical overlap with CHARGE syndrome. Clinical Genetics, 2009, 75, 65-71.	1.0	208
93	A crystallographic snapshot of tyrosine trans -phosphorylation in action. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 19660-19665.	3.3	61
94	Mutations inProkineticin 2andProkineticin receptor 2genes in Human Gonadotrophin-Releasing Hormone Deficiency: Molecular Genetics and Clinical Spectrum. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3551-3559.	1.8	190
95	Relative Roles of Inhibin B and Sex Steroids in the Negative Feedback Regulation of Follicle-Stimulating Hormone in Men across the Full Spectrum of Seminiferous Epithelium Function. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1809-1814.	1.8	40
96	Inhibition of Luteinizing Hormone Secretion by Testosterone in Men Requires Aromatization for Its Pituitary But Not Its Hypothalamic Effects: Evidence from the Tandem Study of Normal and Gonadotropin-Releasing Hormone-Deficient Men. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 784-791.	1.8	119
97	The Relative Role of Gonadal Sex Steroids and Gonadotropin-Releasing Hormone Pulse Frequency in the Regulation of Follicle-Stimulating Hormone Secretion in Men. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 2686-2692.	1.8	55
98	Decreased FGF8 signaling causes deficiency of gonadotropin-releasing hormone in humans and mice. Journal of Clinical Investigation, 2008, 118, 2822-2831.	3.9	348
99	New genes controlling human reproduction and how you find them. Transactions of the American Clinical and Climatological Association, 2008, 119, 29-37; discussion 37-8.	0.9	28
100	Reversal of Idiopathic Hypogonadotropic Hypogonadism. New England Journal of Medicine, 2007, 357, 863-873.	13.9	362
101	Loss-of-function mutation in the prokineticin 2 gene causes Kallmann syndrome and normosmic idiopathic hypogonadotropic hypogonadism. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17447-17452.	3.3	245
102	Acute Sex Steroid Withdrawal Reduces Insulin Sensitivity in Healthy Men with Idiopathic Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 4254-4259.	1.8	208
103	Digenic mutations account for variable phenotypes in idiopathic hypogonadotropic hypogonadism. Journal of Clinical Investigation, 2007, 117, 457-463.	3.9	338
104	Mutations in fibroblast growth factor receptor 1 cause Kallmann syndrome with a wide spectrum of reproductive phenotypes. Molecular and Cellular Endocrinology, 2006, 254-255, 60-69.	1.6	176
105	Testis morphology in patients with idiopathic hypogonadotropic hypogonadism. Human Reproduction, 2006, 21, 1033-1040.	0.4	19
106	Coding sequence analysis of GNRHR and GPR54 in patients with congenital and adult-onset forms of hypogonadotropic hypogonadism. European Journal of Endocrinology, 2006, 155, S3-S10.	1.9	72
107	Mutations in fibroblast growth factor receptor 1 cause both Kallmann syndrome and normosmic idiopathic hypogonadotropic hypogonadism. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 6281-6286.	3.3	225
108	Reversible Kallmann Syndrome, Delayed Puberty, and Isolated Anosmia Occurring in a Single Family with a Mutation in the Fibroblast Growth Factor Receptor 1 Gene. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 1317-1322.	1.8	144

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109	Relationship Between Testosterone Levels, Insulin Sensitivity, and Mitochondrial Function in Men. Diabetes Care, 2005, 28, 1636-1642.	4.3	392
110	Increasing Insulin Resistance Is Associated with a Decrease in Leydig Cell Testosterone Secretion in Men. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 2636-2641.	1.8	424
111	Acute Stress Masking the Biochemical Phenotype of Partial Androgen Insensitivity Syndrome in a Patient with a Novel Mutation in the Androgen Receptor. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 1053-1058.	1.8	4
112	The Role of Prior Pubertal Development, Biochemical Markers of Testicular Maturation, and Genetics in Elucidating the Phenotypic Heterogeneity of Idiopathic Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 152-160.	1.8	175
113	Predictors of Outcome of Long-Term GnRH Therapy in Men with Idiopathic Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4128-4136.	1.8	210
114	The Fertile Eunuch Variant of Idiopathic Hypogonadotropic Hypogonadism: Spontaneous Reversal Associated with a Homozygous Mutation in the Gonadotropin-Releasing Hormone Receptor1. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 2470-2475.	1.8	92
115	Importance of Inhibin B in the Regulation of FSH Secretion in the Human Male. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5541-5546.	1.8	72
116	Prevalence, Phenotypic Spectrum, and Modes of Inheritance of Gonadotropin-Releasing Hormone Receptor Mutations in Idiopathic Hypogonadotropic Hypogonadism¹. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 1580-1588.	1.8	205
117	The Fertile Eunuch Variant of Idiopathic Hypogonadotropic Hypogonadism: Spontaneous Reversal Associated with a Homozygous Mutation in the Gonadotropin-Releasing Hormone Receptor. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 2470-2475.	1.8	73
118	Characteristics of Caucasian type 2 diabetic patients during ketoacidosis and at follow-up. Swiss Medical Weekly, 2000, 130, 576-82.	0.8	23
119	Ketoacidosis During Gestational Diabetes: Case report. Diabetes Care, 1998, 21, 1031-1032.	4.3	7