Nicolas de Roux

List of Publications by Year
in descending order

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47	4,752	257101 24	253896 43
papers	citations	h-index	g-index

50	50	50	3799
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hypogonadotropic hypogonadism due to loss of function of the KiSS1-derived peptide receptor GPR54. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 10972-10976.	3.3	2,094
2	European Consensus Statement on congenital hypogonadotropic hypogonadismâ€"pathogenesis, diagnosis and treatment. Nature Reviews Endocrinology, 2015, 11, 547-564.	4.3	664
3	A Family with Hypogonadotropic Hypogonadism and Mutations in the Gonadotropin-Releasing Hormone Receptor. New England Journal of Medicine, 1997, 337, 1597-1603.	13.9	473
4	Mutations in the maternally imprinted gene MKRN3 are common in familial central precocious puberty. European Journal of Endocrinology, 2016, 174, 1-8.	1.9	164
5	Neuroendocrine Phenotype Analysis in Five Patients with Isolated Hypogonadotropic Hypogonadism due to a L102P Inactivating Mutation of GPR54. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 1137-1144.	1.8	153
6	<scp>IGSF</scp> 10 mutations dysregulate gonadotropinâ€releasing hormone neuronal migration resulting in delayed puberty. EMBO Molecular Medicine, 2016, 8, 626-642.	3.3	109
7	A Novel Loss-of-Function Mutation inGPR54/KISS1RLeads to Hypogonadotropic Hypogonadism in a Highly Consanguineous Family. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E536-E545.	1.8	76
8	GnRH receptor mutations in isolated gonadotropic deficiency. Molecular and Cellular Endocrinology, 2011, 346, 21-28.	1.6	67
9	Haploinsufficiency of Dmxl2, Encoding a Synaptic Protein, Causes Infertility Associated with a Loss of GnRH Neurons in Mouse. PLoS Biology, 2014, 12, e1001952.	2.6	66
10	Mutation Ala171Thr Stabilizes the Gonadotropin-Releasing Hormone Receptor in Its Inactive Conformation, Causing Familial Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 1873-1879.	1.8	56
11	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
12	High tumoral levels of Kiss1 and G-protein-coupled receptor 54 expression are correlated with poor prognosis of estrogen receptor-positive breast tumors. Endocrine-Related Cancer, 2007, 14, 691-702.	1.6	54
13	Linkage and mutational analysis of familial thyroid dysgenesis demonstrate genetic heterogeneity implicating novel genes. European Journal of Human Genetics, 2005, 13, 232-239.	1.4	49
14	Paediatric phenotype of Kallmann syndrome due to mutations of fibroblast growth factor receptor 1 (FGFR1). Molecular and Cellular Endocrinology, 2006, 254-255, 78-83.	1.6	48
15	Clinical and molecular genetics of the human GnRH receptor. Human Reproduction Update, 2003, 9, 523-530.	5.2	46
16	The Incidence of Rett Syndrome in France. Pediatric Neurology, 2006, 34, 372-375.	1.0	44
17	DLK1 Is a Somato-Dendritic Protein Expressed in Hypothalamic Arginine-Vasopressin and Oxytocin Neurons. PLoS ONE, 2012, 7, e36134.	1.1	43
18	TSH receptor mutation V509A causes familial hyperthyroidism by release of interhelical constraints between transmembrane helices TMH3 and TMH5. Journal of Endocrinology, 2005, 186, 377-385.	1.2	42

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19	Negative Fetal FSH/LH Regulation in Late Pregnancy Is Associated with Declined Kisspeptin/KISS1R Expression in the Tuberal Hypothalamus. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2221-E2229.	1.8	41
20	Dental agenesis in Kallmann syndrome individuals with FGFR1 mutations. International Journal of Paediatric Dentistry, 2010, 20, 305-312.	1.0	34
21	Rational Design of Triazololipopeptides Analogs of Kisspeptin Inducing a Long-Lasting Increase of Gonadotropins. Journal of Medicinal Chemistry, 2015, 58, 3459-3470.	2.9	34
22	Molecular Genetics of Isolated Hypogonadotropic Hypogonadism and Kallmann Syndrome. , 2005, 8, 67-80.		33
23	An Inactivating Mutation within the First Extracellular Loop of the Thyrotropin Receptor Impedes Normal Posttranslational Maturation of the Extracellular Domain. Endocrinology, 2009, 150, 1043-1050.	1.4	32
24	GnRH receptor and GPR54 inactivation in isolated gonadotropic deficiency. Best Practice and Research in Clinical Endocrinology and Metabolism, 2006, 20, 515-528.	2.2	30
25	PROKR2 and PROK2 mutations cause isolated congenital anosmia without gonadotropic deficiency. European Journal of Endocrinology, 2013, 168, 31-37.	1.9	27
26	Zero-Length Cross-Linking Reveals that Tight Interactions between the Extracellular and Transmembrane Domains of the Luteinizing Hormone Receptor Persist during Receptor Activation. Molecular Endocrinology, 2005, 19, 2086-2098.	3.7	26
27	DNA polymorphisms of the KiSS1 3â€² Untranslated region interfere with the folding of a G-rich sequence into G-quadruplex. Molecular and Cellular Endocrinology, 2012, 351, 239-248.	1.6	26
28	KISS1 Is Down-Regulated by 17Î²-Estradiol in MDA-MB-231 Cells through a Nonclassical Mechanism and Loss of Ribonucleic Acid Polymerase II Binding at the Proximal Promoter. Endocrinology, 2010, 151, 3764-3772.	1.4	20
29	PRR Repeats in the Intracellular Domain of KISS1R Are Important for Its Export to Cell Membrane. Molecular Endocrinology, 2013, 27, 1004-1014.	3.7	18
30	Spectrum of MECP2 Mutations in Rett Syndrome. Genetic Testing and Molecular Biomarkers, 2002, 6, 1-6.	1.7	17
31	Absence of GPR54 and TACR3 Mutations in Sporadic Cases of Idiopathic Central Precocious Puberty. Hormone Research in Paediatrics, 2014, 81, 177-181.	0.8	14
32	Rabconnectin-3Î± is required for the morphological maturation of GnRH neurons and kisspeptin responsiveness. Scientific Reports, 2017, 7, 42463.	1.6	14
33	High prevalence of syndromic disorders in patients with non-isolated central precocious puberty. European Journal of Endocrinology, 2018, 179, 373-380.	1.9	13
34	Cerebellar hypoplasia with endosteal sclerosis is a POLR3-related disorder. European Journal of Human Genetics, 2017, 25, 1011-1014.	1.4	11
35	Isolated Gonadotropic Deficiency with and without Anosmia: A Developmental Defect or a Neuroendocrine Regulation Abnormality of the Gonadotropic Axis. Hormone Research in Paediatrics, 2005, 64, 48-55.	0.8	10
36	Congenital Hypogonadotropic Hypogonadism: A Trait Shared by Several Complex Neurodevelopmental Disorders. Endocrine Development, 2016, 29, 72-86.	1.3	9

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37	Pituitary-Thyroid Feedback in a Patient with a Sporadic Activating Thyrotropin (TSH) Receptor Mutation: Implication That Thyroid-Secreted Factors Other Than Thyroid Hormones Contribute to Serum TSH Levels. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 2787-2791.	1.8	8
38	Somatostatin Receptors Type 2 and 5 Expression and Localization During Human Pituitary Development. Endocrinology, 2014, 155, 33-39.	1.4	5
39	Complete Kisspeptin Receptor Inactivation Does Not Impede Exogenous GnRH-Induced LH Surge in Humans. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4482-4490.	1.8	5
40	Functional characterization of the novel sequence variant p.S304R in the hinge region of TSHR in a congenital hypothyroidism patients and analogy with other formerly known mutations of this gene portion. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 777-84.	0.4	4
41	Genotype-Phenotype Relationship in Patients and Relatives with <i>SHOX</i> Region Anomalies in the French Population. Hormone Research in Paediatrics, 2016, 86, 309-318.	0.8	4
42	Estrogen alpha receptor inactivation in two sisters: different phenotypic severities for the same pathogenic variant. Journal of Clinical Endocrinology and Metabolism, 2022, , .	1.8	3
43	Gonadotropic Axis Deficiency: A Neurodevelopmental Disorder. Research and Perspectives in Endocrine Interactions, 2015, , 155-162.	0.2	1
44	3Î²-Hydroxysteroid Dehydrogenase Type 2 (3Î²HSD2) Deficiency due to a Novel Compound Heterozygosity of a Missense Mutation (p.Thr259Met) and Frameshift Deletion (p.Lys273ArgFs*7) in an Undervirilized Infant Male with Salt Wasting. Sexual Development, 2022, 16, 64-69.	1.1	1
45	Lack of detection of novel nonsense mutations on exon 3 of hemochromatosis gene in patients with hepatic iron overload. Journal of Hepatology, 2004, 40, 182-183.	1.8	0
46	Neuroendocrine Control of Puberty. , 2016, , 21-30.		0
47	Le contrÃ1le gÃ©nÃ©tique de l'initiation de la pubertÃ©. Bulletin De L'Academie Nationale De Medecine, 2012, 196, 327-343.	0.0	0