Nicolas de Roux

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6484584/publications.pdf

Version: 2024-02-01

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	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
2	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
3	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
4	High prevalence of syndromic disorders in patients with non-isolated central precocious puberty. European Journal of Endocrinology, 2018, 179, 373-380.	1.9	13
5	Rabconnectin-3Î± is required for the morphological maturation of GnRH neurons and kisspeptin responsiveness. Scientific Reports, 2017, 7, 42463.	1.6	14
6	Cerebellar hypoplasia with endosteal sclerosis is a POLR3-related disorder. European Journal of Human Genetics, 2017, 25, 1011-1014.	1.4	11
7	Congenital Hypogonadotropic Hypogonadism: A Trait Shared by Several Complex Neurodevelopmental Disorders. Endocrine Development, 2016, 29, 72-86.	1.3	9
8	<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
9	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
10	Neuroendocrine Control of Puberty. , 2016, , 21-30.		0
11	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
12	Gonadotropic Axis Deficiency: A Neurodevelopmental Disorder. Research and Perspectives in Endocrine Interactions, 2015, , 155-162.	0.2	1
13	European Consensus Statement on congenital hypogonadotropic hypogonadismâ€”pathogenesis, diagnosis and treatment. Nature Reviews Endocrinology, 2015, 11, 547-564.	4.3	664
14	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
15	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
16	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
17	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
18	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

#	Article	IF	Citations
19	Somatostatin Receptors Type 2 and 5 Expression and Localization During Human Pituitary Development. Endocrinology, 2014, 155, 33-39.	1.4	5
20	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
21	PROKR2 and PROK2 mutations cause isolated congenital anosmia without gonadotropic deficiency. European Journal of Endocrinology, 2013, 168, 31-37.	1.9	27
22	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
23	DLK1 Is a Somato-Dendritic Protein Expressed in Hypothalamic Arginine-Vasopressin and Oxytocin Neurons. PLoS ONE, 2012, 7, e36134.	1.1	43
24	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
25	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
26	GnRH receptor mutations in isolated gonadotropic deficiency. Molecular and Cellular Endocrinology, 2011, 346, 21-28.	1.6	67
27	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
28	Dental agenesis in Kallmann syndrome individuals with FGFR1 mutations. International Journal of Paediatric Dentistry, 2010, 20, 305-312.	1.0	34
29	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
30	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
31	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
32	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
33	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
34	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
35	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
36	The Incidence of Rett Syndrome in France. Pediatric Neurology, 2006, 34, 372-375.	1.0	44

#	Article	IF	Citations
37	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
38	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
39	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
40	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
41	Molecular Genetics of Isolated Hypogonadotropic Hypogonadism and Kallmann Syndrome. , 2005, 8, 67-80.		33
42	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
43	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
44	Clinical and molecular genetics of the human GnRH receptor. Human Reproduction Update, 2003, 9, 523-530.	5.2	46
45	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
46	Spectrum of MECP2 Mutations in Rett Syndrome. Genetic Testing and Molecular Biomarkers, 2002, 6, 1-6.	1.7	17
47	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