

Luciani R Carvalho

List of Publications by Year in descending order

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42
papers

1,013
citations

516215

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docs citations

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#	ARTICLE	IF	CITATIONS
1	Rederivation of a mutant line (prop 1) of zebrafish <i>Danio rerio</i> infected with <i>Pseudoloma neurophilia</i> using in vitro fertilization with eggs from pathogen-free wild-type (AB) females and sperm from prop 1 males. <i>Journal of Fish Diseases</i> , 2022, 45, 35-39.	0.9	3
2	Cushing disease due to a somatic USP8 mutation in a patient with evolving pituitary hormone deficiencies due to a germline GH1 splicing variant. <i>Archives of Endocrinology and Metabolism</i> , 2022, , .	0.3	2
3	Novel OTX2 loss of function variant associated with congenital hypopituitarism without eye abnormalities. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2022, .	0.4	0
4	Central adrenal insufficiency: who, when, and how? From the evidence to the controversies – an exploratory review. <i>Archives of Endocrinology and Metabolism</i> , 2022, , .	0.3	2
5	Allelic Variants in Established Hypopituitarism Genes Expand Our Knowledge of the Phenotypic Spectrum. <i>Genes</i> , 2021, 12, 1128.	1.0	0
6	Metabolomics as a potential tool for the diagnosis of growth hormone deficiency (GHD): a review. <i>Archives of Endocrinology and Metabolism</i> , 2020, 64, 654-663.	0.3	1
7	Combined pituitary hormone deficiency caused by PROP1 mutations: update 20 years post-discovery. <i>Archives of Endocrinology and Metabolism</i> , 2019, 63, 167-174.	0.3	23
8	Genetic Evidence of the Association of DEAH-Box Helicase 37 Defects With 46,XY Gonadal Dysgenesis Spectrum. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 5923-5934.	1.8	26
9	An Easy Method for Cryopreservation of Zebrafish (<i>Danio rerio</i>) Sperm. <i>Zebrafish</i> , 2019, 16, 321-323.	0.5	3
10	Mobile DNA in Endocrinology: LINE-1 Retrotransposon Causing Partial Androgen Insensitivity Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 6385-6390.	1.8	10
11	OR06-6 Whole-Exome Sequencing of Patients with Pituitary Stalk Interruption Syndrome (PSIS) Reveals Probably Pathogenic Variants in Novel Candidate Genes.. <i>Journal of the Endocrine Society</i> , 2019, 3, .	0.1	0
12	Pathogenic copy number variants in patients with congenital hypopituitarism associated with complex phenotypes. <i>Clinical Endocrinology</i> , 2018, 88, 425-431.	1.2	11
13	A recurrent synonymous mutation in the human androgen receptor gene causing complete androgen insensitivity syndrome. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2017, 174, 14-16.	1.2	16
14	Molecular analysis of brazilian patients with combined pituitary hormone deficiency and orthotopic posterior pituitary lobe reveals eight different <i>PROP1</i> alterations with three novel mutations. <i>Clinical Endocrinology</i> , 2017, 87, 725-732.	1.2	13
15	Differential Expression of Stem Cell Markers in Human Adamantinomatous Craniopharyngioma and Pituitary Adenoma. <i>Neuroendocrinology</i> , 2017, 104, 183-193.	1.2	19
16	Successful Pregnancies After Adequate Hormonal Replacement in Patients With Combined Pituitary Hormone Deficiencies. <i>Journal of the Endocrine Society</i> , 2017, 1, 1322-1330.	0.1	14
17	Growth hormone deficiency with advanced bone age: phenotypic interaction between GHRH receptor and CYP21A2 mutations diagnosed by sanger and whole exome sequencing. <i>Archives of Endocrinology and Metabolism</i> , 2017, 61, 633-636.	0.3	4
18	<i>HESX1</i> mutations in patients with congenital hypopituitarism: variable phenotypes with the same genotype. <i>Clinical Endocrinology</i> , 2016, 85, 408-414.	1.2	24

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19	A homozygous point mutation in the GH1 promoter (c.-223C>T) leads to reduced GH1 expression in siblings with isolated GH deficiency (IGHD). <i>European Journal of Endocrinology</i> , 2016, 175, K7-K15.	1.9	5
20	Ketoconazole Treatment Decreases the Viability of Immortalized Pituitary Cell Lines Associated with an Increased Expression of Apoptosis-Related Genes and Cell Cycle Inhibitors. <i>Journal of Neuroendocrinology</i> , 2015, 27, 616-623.	1.2	5
21	Frequent development of combined pituitary hormone deficiency in patients initially diagnosed as isolated growth hormone deficiency: a long term follow-up of patients from a single center. <i>Pituitary</i> , 2015, 18, 561-567.	1.6	30
22	Role of GLI2 in hypopituitarism phenotype. <i>Journal of Molecular Endocrinology</i> , 2015, 54, R141-R150.	1.1	50
23	FGFR1 and PROKR2 rare variants found in patients with combined pituitary hormone deficiencies. <i>Endocrine Connections</i> , 2015, 4, 100-107.	0.8	34
24	Apoptosis: its role in pituitary development and neoplastic pituitary tissue. <i>Pituitary</i> , 2014, 17, 157-162.	1.6	12
25	Autosomal recessive form of isolated growth hormone deficiency is more frequent than the autosomal dominant form in a Brazilian cohort. <i>Growth Hormone and IGF Research</i> , 2014, 24, 180-186.	0.5	5
26	Relatively high frequency of non-synonymous <i>GLI2</i> variants in patients with congenital hypopituitarism without holoprosencephaly. <i>Clinical Endocrinology</i> , 2013, 78, 551-557.	1.2	33
27	PROP1 overexpression in corticotrophinomas: evidence for the role of PROP1 in the maintenance of cells committed to corticotrophic differentiation. <i>Clinics</i> , 2013, 68, 887-891.	0.6	9
28	Absence of GH-Releasing Hormone (GHRH) Mutations in Selected Patients with Isolated GH Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E1457-E1460.	1.8	17
29	PROP1 and CTNNB1 expression in adamantinomatous craniopharyngiomas with or without β -catenin mutations. <i>Clinics</i> , 2011, 66, 1849-54.	0.6	13
30	Novel Heterozygous Nonsense GLI2 Mutations in Patients with Hypopituitarism and Ectopic Posterior Pituitary Lobe without Holoprosencephaly. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, E384-E391.	1.8	91
31	Corepressors TLE1 and TLE3 Interact with HESX1 and PROP1. <i>Molecular Endocrinology</i> , 2010, 24, 754-765.	3.7	23
32	Molecular mechanisms of pituitary organogenesis: In search of novel regulatory genes. <i>Molecular and Cellular Endocrinology</i> , 2010, 323, 4-19.	1.6	140
33	Analysis of Craniofacial and Extremity Growth in Patients with Growth Hormone Deficiency during Growth Hormone Therapy. <i>Hormone Research</i> , 2009, 71, 173-177.	1.8	12
34	Expression profiles of the glucose-dependent insulinotropic peptide receptor and LHCGR in sporadic adrenocortical tumors. <i>Journal of Endocrinology</i> , 2009, 200, 167-175.	1.2	5
35	Comparison between weight-based and IGF-I-based growth hormone (GH) dosing in the treatment of children with GH deficiency and influence of exon 3 deleted GH receptor variant. <i>Growth Hormone and IGF Research</i> , 2009, 19, 179-186.	0.5	16
36	Tall stature and poor breast development after estrogen replacement in a hypergonadotrophic hypogonadic patient with a 45,X/46,X,der(X) karyotype with SHOX gene overdosage. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , 2008, 52, 1282-1287.	1.3	9

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37	Combined pituitary hormone deficiency (CPHD) due to a complete PROP1 deletion. Clinical Endocrinology, 2006, 65, 294-300.	1.2	35
38	Hormonal, pituitary magnetic resonance, LHX4 and HESX1 evaluation in patients with hypopituitarism and ectopic posterior pituitary lobe. Clinical Endocrinology, 2006, 66, 061107003613003-???.	1.2	32
39	Growth Hormone (GH) Pharmacogenetics: Influence of GH Receptor Exon 3 Retention or Deletion on First-Year Growth Response and Final Height in Patients with Severe GH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1076-1080.	1.8	136
40	Craniofacial features with growth hormone treatment. Journal of Pediatrics, 2005, 146, 295.	0.9	1
41	Acromegalic features in growth hormone (GH)-deficient patients after long-term GH therapy. Clinical Endocrinology, 2003, 59, 788-792.	1.2	18
42	A homozygous mutation in HESX1 is associated with evolving hypopituitarism due to impaired repressor-corepressor interaction. Journal of Clinical Investigation, 2003, 112, 1192-1201.	3.9	110