

Lucas Santana

List of Publications by Year in descending order

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Version: 2024-02-01

20
papers

170
citations

1040056

9
h-index

1199594

12
g-index

21
all docs

21
docs citations

21
times ranked

309
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>PDX1</i> and <i>MODY</i> and dorsal pancreatic agenesis: New phenotype of a rare disease. <i>Clinical Genetics</i> , 2018, 93, 382-386.	2.0	25
2	Targeted sequencing identifies novel variants in common and rare <i>MODY</i> genes. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e962.	1.2	24
3	Clinical application of ACMG-AMP guidelines in <i>HNF1A</i> and <i>GCK</i> variants in a cohort of <i>MODY</i> families. <i>Clinical Genetics</i> , 2017, 92, 388-396.	2.0	19
4	Phosphodiesterase 2A and 3B variants are associated with primary aldosteronism. <i>Endocrine-Related Cancer</i> , 2021, 28, 1-13.	3.1	17
5	Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. <i>Human Reproduction</i> , 2021, 36, 506-518.	0.9	16
6	Germline mutation landscape of multiple endocrine neoplasia type 1 using full gene next-generation sequencing. <i>European Journal of Endocrinology</i> , 2018, 179, 391-407.	3.7	14
7	Homozygous and Heterozygous Nuclear Lamin A p.R582C Mutation: Different Lipodystrophic Phenotypes in the Same Kindred. <i>Frontiers in Endocrinology</i> , 2018, 9, 458.	3.5	13
8	Comprehensive Genetic Analysis of 128 Candidate Genes in a Cohort With Idiopathic, Severe, or Familial Osteoporosis. <i>Journal of the Endocrine Society</i> , 2020, 4, bvaa148.	0.2	11
9	Genetic diagnosis of congenital hypopituitarism by a target gene panel: novel pathogenic variants in <i>GLI2</i> , <i>OTX2</i> and <i>GHRHR</i> . <i>Endocrine Connections</i> , 2019, 8, 590-595.	1.9	10
10	Evaluation of <i>SHOX</i> defects in the era of next-generation sequencing. <i>Clinical Genetics</i> , 2019, 96, 261-265.	2.0	9
11	Searching for mutations in the <i>HNF1B</i> gene in a Brazilian cohort with renal cysts and hyperglycemia. <i>Archives of Endocrinology and Metabolism</i> , 2019, 63, 250-257.	0.6	6
12	Pathogenesis of Primary Aldosteronism: Impact on Clinical Outcome. <i>Frontiers in Endocrinology</i> , 0, 13, .	3.5	4
13	Clonagem molecular do oncogene <i>EZH2</i> de leucemia mieloide crônica e perspectivas terapêuticas. <i>Mundo Da Saude</i> , 2015, 39, 307-315.	0.1	1
14	Next-generation sequencing in Brazilian <i>MODY</i> patients: a pilot study. <i>Diabetology and Metabolic Syndrome</i> , 2015, 7, .	2.7	0
15	<i>MODY</i> screening: a new center for molecular genetic diagnosis in Brazil. <i>Diabetology and Metabolic Syndrome</i> , 2015, 7, A213.	2.7	0
16	Detection of congenital generalized lipodystrophy mutations by next-generation sequencing: time for a new approach. <i>Diabetology and Metabolic Syndrome</i> , 2015, 7, .	2.7	0
17	Comparative Analysis of Different International Criteria (ACMG-AMP vs. TENGEN) Applied to Classification of Missense Germline Allelic Variants in Patients With Multiple Endocrine Neoplasia Type 1 or Suspected to this Syndrome. <i>Journal of the Endocrine Society</i> , 2021, 5, A1014-A1014.	0.2	0
18	Comprehensive Analysis of Clinical Features in Index Cases With Multiple Endocrine Neoplasia Type 1 Refine the Risk Rate for Detection of Mutation Distinguishing Negative-Mutation (Phenocopies) and Positive-Mutation Cases. <i>Journal of the Endocrine Society</i> , 2021, 5, A1014-A1015.	0.2	0

#	ARTICLE	IF	CITATIONS
19	SUN-034 Genetic Diagnosis of Congenital Isolated or Combined Growth Hormone Deficiency by Massive Parallel Sequencing Using a Target Gene Panel. Journal of the Endocrine Society, 2019, 3, .	0.2	0
20	Targeted massively parallel sequencing for congenital generalized lipodystrophy. Archives of Endocrinology and Metabolism, 2020, 64, 559-566.	0.6	0