CLAUDIA GABRIELA GONZAGA JAUREGUI

Datos Generales

Nombre: CLAUDIA GABRIELA GONZAGA JAUREGUI

Máximo nivel de estudios: DOCTORADO

Antigüedad académica en la UNAM: 4 años

Nombramientos

Vigente: INVESTIGADOR ASOCIADO C TC No Definitivo
Laboratorio Internacional de Investigación Sobre el Genoma Humano
Desde 01-10-2020

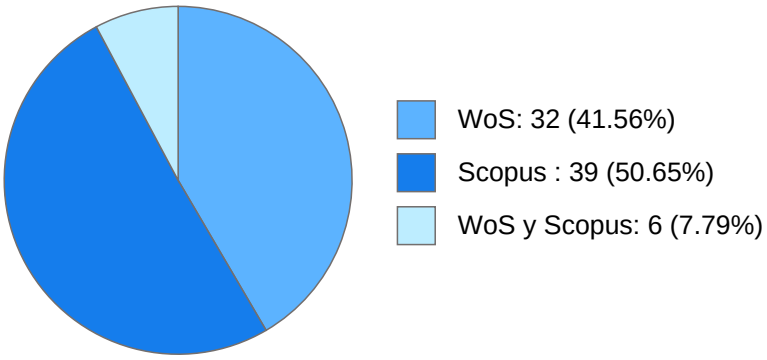
Estímulos, programas, premios y reconocimientos

EQUIVALENCIA PRIDE B 2020 - 2024

CLAUDIA GABRIELA GONZAGA JAUREGUI

DOCUMENTOS EN REVISTAS

Histórico de Documentos



#	Título	Autores	Revista	Año
1	Genome-wide maps of highly- similar intrachromosomal repeats that can mediate ectopic recombination in three human genome assemblies	CLAUDIA GABRIELA GONZAGA JAUREGUI Luis Fernandez-Luna Carlos Aguilar-Perez et al.	Human Genetics And Genomics Advances	2025
2	Design and implementation of an action plan for justice, equity, diversity, and inclusion within the Clinical Genome Resource	CLAUDIA GABRIELA GONZAGA JAUREGUI Popejoy A.B. Ritter D.I. et al.	AMERICAN JOURNAL OF HUMAN GENETICS	2025
3	Drug?device combinations in rare diseases: Challenges and opportunities	CLAUDIA GABRIELA GONZAGA JAUREGUI Tataru E.A. Dooms M. et al.	DRUG DISCOVERY TODAY	2025
4	A rare variant in GPR156 associated with depression in a Mennonite pedigree causes habenula hyperactivity and stress sensitivity in mice	CLAUDIA GABRIELA GONZAGA JAUREGUI CRISTOPHER VINCENT VAN HOUT Miller B.R. et al.	PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA	2025
5	Population-Specific Differences in Pathogenic Variants of Genes Associated with Monogenic Parkinson's Disease	CLAUDIA GABRIELA GONZAGA JAUREGUI Victor Flores-Ocampo Amanda Wei-Yin Lim et al.	GENES	2025

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6	Newborn screening in Mexico and Latin America: present and future	CLAUDIA GABRIELA GONZAGA JAUREGUI JUANA INES NAVARRETE MARTINEZ Moreno-Salgado R. et al.	Rare Disease And Orphan Drugs Journal	2024
7	ERCAL, a regional initiative for rare diseases in Latin America and the Caribbean	CLAUDIA GABRIELA GONZAGA JAUREGUI Salazar C. Macdonald J. et al.	Rare Disease And Orphan Drugs Journal	2024
8	Rare Variant in MRC2 Associated With Familial Supraventricular Tachycardia and Wolff-Parkinson-White Syndrome	CLAUDIA GABRIELA GONZAGA JAUREGUI Adam S. Potter Christina Y. Miyake et al.	CIRCULATION-GENOMIC AND PRECISION MEDICINE	2024
9	Pushing the boundaries of rare disease diagnostics with the help of the first Undiagnosed Hackathon	CLAUDIA GABRIELA GONZAGA JAUREGUI Delgado-Vega A.M. Cederroth H. et al.	NATURE GENETICS	2024
10	High-coverage nanopore sequencing of samples from the 1000 Genomes Project to build a comprehensive catalog of human genetic variation	CLAUDIA GABRIELA GONZAGA JAUREGUI Gustafson J.A. Gibson S.B. et al.	GENOME RESEARCH	2024
11	Unmet needs in countries participating in the undiagnosed diseases network international: an international survey considering national health care and economic indicators	CLAUDIA GABRIELA GONZAGA JAUREGUI Sciascia S. Roccatello D. et al.	Frontiers In Public Health	2023
12	Hemizygous variants in protein phosphatase 1 regulatory subunit 3F (PPP1R3F) are associated with a neurodevelopmental disorder characterized by developmental delay, intellectual disability and autistic features	CLAUDIA GABRIELA GONZAGA JAUREGUI Zhigang Liu Baozhong Xin et al.	HUMAN MOLECULAR GENETICS	2023
13	Comprehensive Genetic Analysis of Druze Provides Insights into Carrier Screening	CLAUDIA GABRIELA GONZAGA JAUREGUI Eden Avnat Guy Shapira et al.	GENES	2023
14	Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International	CLAUDIA GABRIELA GONZAGA JAUREGUI Taruscio D. Salvatore M. et al.	Frontiers In Public Health	2023
15	Addressing the challenges of polygenic scores in human genetic research	CLAUDIA GABRIELA GONZAGA JAUREGUI John Novembre Catherine Stein et al.	AMERICAN JOURNAL OF HUMAN GENETICS	2022
16	Clinical validity assessment of genes frequently tested on intellectual disability/autism sequencing panels	CLAUDIA GABRIELA GONZAGA JAUREGUI Erin Rooney Riggs I Bingaman et al.	GENETICS IN MEDICINE	2022

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17	Editorial: Chromosome structural variants: Epidemiology, identification and contribution to human diseases	CLAUDIA GABRIELA GONZAGA JAUREGUI Zirui Dong Dezso David et al.	Frontiers in Genetics	2022
18	Pathogenic variants in SLF2 and SMC5 cause segmented chromosomes and mosaic variegated hyperploidy	CLAUDIA GABRIELA GONZAGA JAUREGUI Grange L.J. Reynolds J.J. et al.	NATURE COMMUNICATIONS	2022
19	Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America	CLAUDIA GABRIELA GONZAGA JAUREGUI Roberto Giugliani Silvia Castillo Taucher et al.	Frontiers in Genetics	2022
20	Biallelic truncating variants in the muscular A-type lamin-interacting protein (MLIP) gene cause myopathy with hyperCKemia	CRISTOPHER VINCENT VAN HOUT CLAUDIA GABRIELA GONZAGA JAUREGUI Liat Salzer-Sheelo et al.	EUROPEAN JOURNAL OF NEUROLOGY	2022
21	Mucus sialylation determines intestinal host-commensal homeostasis	CLAUDIA GABRIELA GONZAGA JAUREGUI Yikun Yao Girak Kim et al.	Cell	2022
22	Clinical characterization of familial hypercholesterolemia due to an amish founder mutation in Apolipoprotein B	CLAUDIA GABRIELA GONZAGA JAUREGUI Williams K.B. Horst M. et al.	BMC CARDIOVASCULAR DISORDERS	2022
23	Correction to: Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension (Genome Medicine, (2019), 11, 1, (69), 10.1186/s13073-019-0685-z)	CLAUDIA GABRIELA GONZAGA JAUREGUI Zhu N. Pauciulo M.W. et al.	GENOME MEDICINE	2022
24	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity	CLAUDIA GABRIELA GONZAGA JAUREGUI Kurolap A. Kreuder F. et al.	AMERICAN JOURNAL OF HUMAN GENETICS	2022
25	A de novo paradigm for male infertility	CLAUDIA GABRIELA GONZAGA JAUREGUI Oud M.S. Smits R.M. et al.	NATURE COMMUNICATIONS	2022
26	NPRL3 loss alters neuronal morphology, mTOR localization, cortical lamination and seizure threshold	CLAUDIA GABRIELA GONZAGA JAUREGUI Iffland P.H. Ii et al.	Brain	2022
27	Assessing structural variation in a personal genome-towards a human reference diploid genome	CLAUDIA GABRIELA GONZAGA JAUREGUI English A.C. Salerno W.J. et al.	Bmc Genomics	2015
28	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy	CLAUDIA GABRIELA GONZAGA JAUREGUI Harel T. Gambin T. et al.	CELL REPORTS	2015

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29	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease	CLAUDIA GABRIELA GONZAGA JAUREGUI Karaca E. Harel T. et al.	Neuron	2015
30	NR2F1 mutations cause optic atrophy with intellectual disability	CLAUDIA GABRIELA GONZAGA JAUREGUI Bosch D.G.M. Boonstra F.N. et al.	AMERICAN JOURNAL OF HUMAN GENETICS	2014
31	Erratum: Exonic duplication CNV of NDRG1 associated with autosomal-recessive HMSN-Lom/CMT4D (Genetics in Medicine (2013) DOI:10.1038/gim.2013.155)	CLAUDIA GABRIELA GONZAGA JAUREGUI Okamoto Y. Goksungur M.T. et al.	GENETICS IN MEDICINE	2014
32	Human CLP1 mutations alter tRNA biogenesis, Affecting both peripheral and central nervous system function	CLAUDIA GABRIELA GONZAGA JAUREGUI Karaca E. Weitzer S. et al.	Cell	2014
33	Exonic duplication CNV of NDRG1 associated with autosomal-recessive HMSN-Lom/CMT4D	CLAUDIA GABRIELA GONZAGA JAUREGUI Okamoto Y. Goksungur M.T. et al.	GENETICS IN MEDICINE	2014
34	Clinical utility of whole-exome sequencing in rare diseases: Galactosialidosis	CLAUDIA GABRIELA GONZAGA JAUREGUI Prada C.E. Tannenbaum R. et al.	EUROPEAN JOURNAL OF MEDICAL GENETICS	2014
35	Monoallelic and biallelic mutations in MAB21L2 cause a spectrum of major eye malformations	CLAUDIA GABRIELA GONZAGA JAUREGUI Rainger J. Pehlivan D. et al.	AMERICAN JOURNAL OF HUMAN GENETICS	2014
36	Mutations in VRK1 associated with complex motor and sensory axonal neuropathy plus microcephaly	CLAUDIA GABRIELA GONZAGA JAUREGUI Lotze T. Jamal L. et al.	JAMA NEUROLOGY	2013
37	Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis	CLAUDIA GABRIELA GONZAGA JAUREGUI Dittwald P. Gambin T. et al.	HUMAN MUTATION	2013
38	Human genome sequencing in health and disease	CLAUDIA GABRIELA GONZAGA JAUREGUI Lupski J.R. Gibbs R.A.	ANNU REV MED	2012
39	Identical repeated backbone of the human genome	DELFINO GARCIA ALONSO David Valle Garcia Karla F. Meza Sosa et al.	Bmc Genomics	2010
40	High-Resolution Copy-Number Variation Map Reflects Human Olfactory Receptor Diversity and Evolution	Hasin, Yehudit Olender, Tsviya Khen, Miriam et al.	PLOS GENETICS	2008

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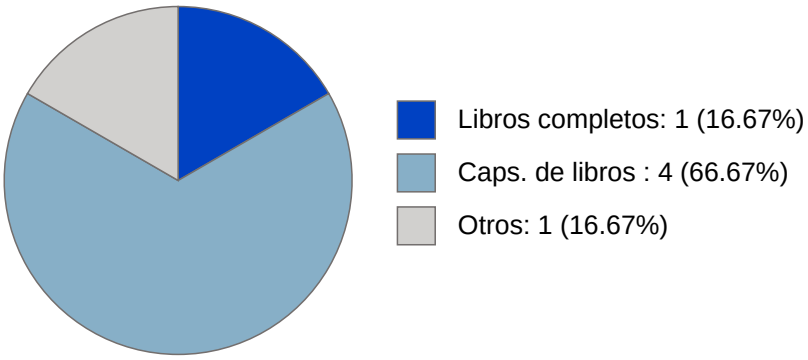
LIBROS Y CAPITULOS CON ISBN

Obras con registro ISBN



#	Título	Autores	Alcance	Año	ISBN
1	Preface	CLAUDIA GABRIELA GONZAGA JAUREGUI Lupski J.R.	Editorial Material	2021	9780128201404
2	Genomic sequencing of rare diseases	CLAUDIA GABRIELA GONZAGA JAUREGUI Zepeda Mendoza C.J.	Capítulo de un Libro	2021	9780128201404
3	Challenges and opportunities in rare diseases research	CLAUDIA GABRIELA GONZAGA JAUREGUI	Capítulo de un Libro	2021	9780128201404
4	Genomics of Rare Diseases: Understanding Disease Genetics Using Genomic Approaches	CLAUDIA GABRIELA GONZAGA JAUREGUI Lupski J.R.	Libro Completo	2021	9780128201404
5	Genomic disorders in the genomics era	CLAUDIA GABRIELA GONZAGA JAUREGUI Zepeda Mendoza C.J.	Capítulo de un Libro	2021	9780128201404
6	Dominant and sporadic de novo disorders	CLAUDIA GABRIELA GONZAGA JAUREGUI El Hayek L. Chahrour M.	Capítulo de un Libro	2021	9780128201404

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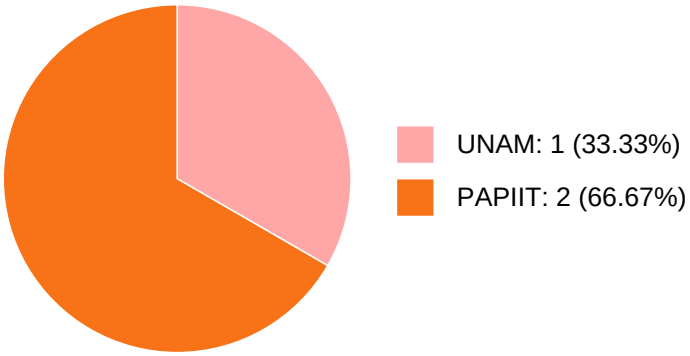
PARTICIPACIÓN EN PROYECTOS

Histórico de participación en proyectos



#	Nombre	Participantes	Fuente	Fecha inicio	Fecha fin
1	Genómica de enfermedades mendelianas.	CLAUDIA GABRIELA GONZAGA JAUREGUI	Presupuesto de la UNAM asignado a la Dependencia	01-08-2020	31-12-2023
2	Secuenciación genómica de pacientes con desórdenes genéticos del Neurodesarrollo	CLAUDIA GABRIELA GONZAGA JAUREGUI	Recursos PAPIIT	01-01-2022	29-03-2024
3	Análisis y modelado funcional de genes asociados a desórdenes genéticos del neurodesarrollo	CLAUDIA GABRIELA GONZAGA JAUREGUI	Recursos PAPIIT	01-01-2024	31-12-2025

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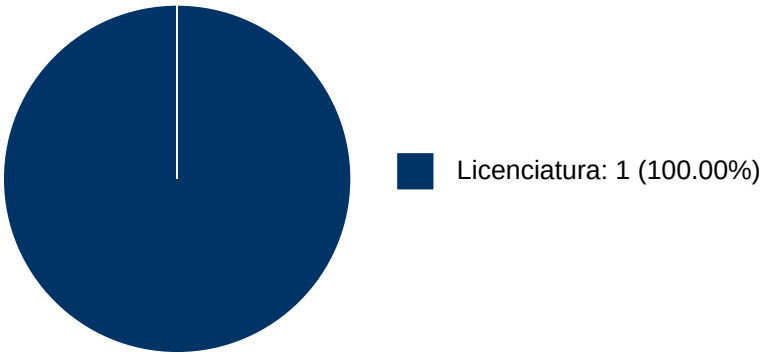
PARTICIPACIÓN EN TESIS

Histórico de Colaboraciones en Tesis

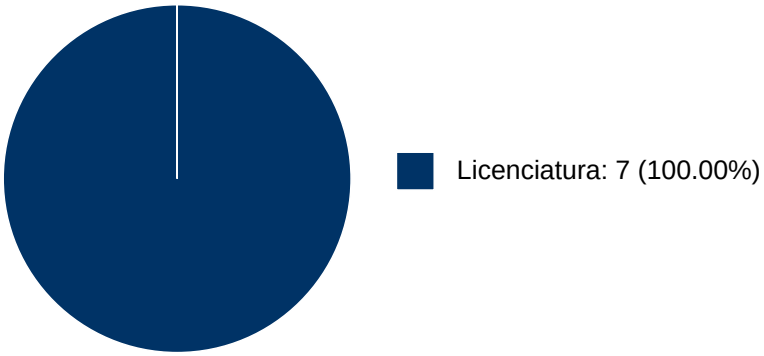


#	Título del documento	Tipo de Tesis	Sinodales	Autores	Entidad	Año
1	Establishing an olfactory classical conditioning task in zebrafish larvae	Tesis de Licenciatura	CLAUDIA GABRIELA GONZAGA JAUREGUI,	Aguilar Pérez, Carlos Gabriel,	Consejo Técnico y Coordinación de la Investigación Científica, Laboratorio Internacional de Investigación Sobre el Genoma Humano,	2022

CLAUDIA GABRIELA GONZAGA JAUREGUI

DOCENCIA IMPARTIDA

Histórico de docencia



#	Nivel titulación	Asignatura	Entidad	Alumnos	Semestre
1	Licenciatura	GENOMICA HUMANA	ESCUELA NACIONAL DE ESTUDIOS SUPERIORES, UNIDAD JURIQUILLA, QRTO.	11	2024-2
2	Licenciatura	GENOMICA INTEGRATIVA 1	ESCUELA NACIONAL DE ESTUDIOS SUPERIORES, UNIDAD JURIQUILLA, QRTO.	12	2024-1
3	Licenciatura	GENOMICA HUMANA	ESCUELA NACIONAL DE ESTUDIOS SUPERIORES, UNIDAD JURIQUILLA, QRTO.	12	2023-2
4	Licenciatura	GENOMICA INTEGRATIVA 1	ESCUELA NACIONAL DE ESTUDIOS SUPERIORES, UNIDAD JURIQUILLA, QRTO.	7	2023-1
5	Licenciatura	GENOMICA INTEGRATIVA 1	ESCUELA NACIONAL DE ESTUDIOS SUPERIORES, UNIDAD JURIQUILLA, QRTO.	14	2022-1
6	Licenciatura	GENOMICA INTEGRATIVA 3	ESCUELA NACIONAL DE ESTUDIOS SUPERIORES, UNIDAD JURIQUILLA, QRTO.	6	2021-2
7	Licenciatura	GENOMICA HUMANA	ESCUELA NACIONAL DE ESTUDIOS SUPERIORES, UNIDAD JURIQUILLA, QRTO.	14	2021-2

CLAUDIA GABRIELA GONZAGA JAUREGUI

PATENTES

No se encuentran registros en la base de datos de patentes asociados a:

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FUENTES DE INFORMACIÓN

Internos

#	Información	Fuente	Sistema	Periodo
1	Grupos ordinarios y resumen de historias académicas	DGAE	SIAE	2008-2025
2	Nombramientos, datos generales, estímulos, premios y reconocimientos	DGAPA	RUPA	2008-2025
3	Producción Académica	CH	Humanindex	2008-2021
4	Producción Académica	CIC	SCIC	2000-2017
5	Proyectos	DGPO	SISEPRO	2018-2022
6	Tesis	DGB	TESIUNAM	2008-2025
7	Tutorías en Posgrado	CGEP	SIIPosgrado	2008-2021

Externos

#	Información	Fuente	Sistema	Periodo
8	Documentos Indexados	Elsevier	Scopus	2008-2025
9	Documentos Indexados	Thomson Reuters	WoS	2008-2025
10	Obras con registro ISBN	INDAUTOR	Agencia ISBN	2008-2025
11	Patentes	IMPI	SIGA	2008-2024