DÍAS ACADÉMICOS LIIGH 2DA EDICIÓN

19 y 20 de Febrero del 2024
Centro Académico Cultural, UNAM
Campus Juriquilla

LIIGH-UNAM

UNAM JURIQUILLA

CAC CENTRO ACADÉMICO CULTURAL

Índice

Comité organizador

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Emmanuel Hernández Sánchez
Federico Sánchez Quinto
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Maria Fernanda García Rodríguez
Maria Fernanda Requena Romo
María José Rodríguez Barrera
Mariana Gómez Schiavon
Sur Herrera Paredes

Objetivo de los días académicos

La segunda edición de los Días Académicos (19 y 20 de febrero de 2024) tiene el objetivo de difundir las investigaciones realizadas en el Laboratorio Internacional de Investigación sobre el Genoma Humano (LIIGH) de la UNAM.

Durante estos días académicos, se imparten conferencias representativas de todos los grupos de investigación activos de nuestra entidad, abordando los avances logrados en sus respectivas áreas. Las áreas de investigación activas en el LIIGH incluyen: genómica de poblaciones, genética del cáncer, paleogenómica, biología de sistemas, medicina de precisión, estadística, bioinformática, ecología y evolución. También se cuenta con dos sesiones de pósters académicos elaborados por estudiantes de todos los niveles, así como posdoctorantes que realizan actividades académicas en el LIIGH.

Comunidad LIIGH Días Académicos 2023.

Programa de pláticas

HORA	LUNES 19
10:00 hrs	**Bienvenida**
	Dra. María del Carmen Ávila Arcos
10:40 hrs	**Conferencia magistral**
	Dr. Federico Sánchez Quinto "*Paleogenómica de la Megafauna y el Paleoambiente de la Cuenca de México*"
11:25 hrs	**LIIGH-3MPC**
11:35 hrs	Pósters y café
	Regulatory Genomics and Bioinformatics Lab
13:05 hrs 13:20 hrs	**Sofia Guadalupe Salazar Magaña** "*Studying the Systemic Lupus Erythematosus expression and contextualizing it in a Mexican cohort*" **Evelia Lorena Coss-Navarrete** "*Project JAGUAR:mapping immune cell diversity across Latin America*"
13:35 hrs	Lonche
	Cancer Genetics and Bioinformatics Lab
15:00 hrs 15:10 hrs 15:20 hrs	**Patricia Basurto Lozada** "*Genomic subtypes of acral lentiginous melanoma tumors associate with clinical outcomes*" **Martha Estefania Vázquez Cruz** *"Transcriptional and spatial protein profiling of acral melanoma tumours from mexican patients"* **Felipe de Jesús Castañeda Córdova** *"The transcriptomic landscape of the fibrotic liver under a time-restricted feeding regime"*
	Ecology and Evolution Lab
15:30 hrs	**Dr. Sur Herrera Paredes** *"Comunidades sintéticas para diseccionar propiedades complejas"*
	Paleogenomics and Evolutionary Biology Lab
16:00 hrs 16:10 hrs 16:20 hrs	**Alan Vladimir Godinez Plascencia** *"Evaluating in silico HLA typing of low-coverage, shotgun sequencing aDNA data"* **Eduardo Alejandro Arrieta Donato** *"Evaluación de imputación en indígenas mexicanos usando paneles de referencia con diferente ancestría"* **Leonardo Yair Correa Mendoza** *"Demographic origin of individuals found in the Sa Galera sanctuary (Spain) from paleogenomic data"*
	Evolutionary Systems Biology Lab
16:30 hrs 16:45 hrs	**Emmanuel Hernández Sánchez** *"Dinámica evolutiva de un modelo del reloj circadiano de Neurospora crassa"* **Manuel Eduardo Hernández García** *"Cuantificando el impacto de las fluctuaciones extrínsecas en el sistema p53"*
17:00 hrs	**Fín del primer día**

HORA	MARTES 20
	Genome Evolution Lab
10:00 hrs 10:10 hrs	**Samuel Quiñones Galeana** "*Exploring Agave Yeast Biodiversity for Bioethanol Production*" **Mariana Guadalupe Guerrero Osornio** "*Time-lapse of the microbial composition during agave fermentation using Meta-HiC*"
10:30 hrs	**LIIGH-3MPC**
	Population and Evolutionary Genomics Lab
10:40 hrs 10:55 hrs	**Laura Carrillo Olivas** "*Exploring Ancient Pathogens in Colonial Mexico City through Paleogenomics*" **Miguel Alejandro Navarro** "*Paleoproteomics and Ancient Pathogen Identification*"
11:10 hrs	**LIIGH-3MPC**
	Mendelian Genomics and Precision Health Lab
11:20 hrs 11:30 hrs 11:40 hrs	**Tania Sepúlveda Morales** "*Estudio de percepciones de la población general en México acerca de las enfermedades raras*" **César Enrique Calvo Aspiros** "*Perspectivas sobre el diagnóstico: Registro Mexicano de Enfermedades Raras*" **Issis Abril Pérez Alvarado** *"Identification of Genomic Variants Associated with Hereditary Cancer in the Mexican Population"*
11:50 hrs	Pósters y café
	Statistical Genomics and Population Health Lab
13:20 hrs 13:35 hrs	**Leonardo Hernandez Delgado** "*Unraveling the genetic basis of Keloids in African Yoruba Individuals*" **Dr. Cris Van Hout** *"Ongoing projects and early ideas in the Statistical Genomics and Population Health group"*
13:50 hrs	**LIIGH-3MPC**
	Computational Population Genetics
14:00 hrs 14:10 hrs 14:20 hrs	**Valeria Alejandra Añorve Garibay** *"The contribution of archaic introgression to the heritability of complex traits in Mexico"* **Héctor Alessandro López Hernández** *"Genética de Poblaciones Espacio-Temporal"* **Alan Raymundo Izarraras Gomez** *"Inferring the impact of natural selection across the genome using local genealogies"*
14:30 hrs	**Clausura**
15:00 hrs	**Evento Social**

Programa de Pósters

Lunes 19

NÚMERO	NOMBRE
1	Diego Morales Martinez
3	Marlon Aldair Arciniega Sánchez
5	Mariana Cuevas Bravo & Cassidy Guadalupe Luna Ruíz
7	Walter Nicolás Ortega
9	Ian Manuel Espinosa
11	Itzy Daniela Pérez Alvarado
13	Bárbara Beatriz Moguel Rodríguez
17	Luis Miguel Pedraza Meza
19	Maria Fernanda Talavera Cruz
21	Maritrini Colón-González
23	Natalia Estrada Hernández
25	Rigoberto Padilla Bustos
27	Christian Molina Aguilar
29	María Fernanda Requena Romo
33	Emiliano Sánchez Escalante
35	Kenya Lizbeth Contreras Ramírez

Martes 20

NÚMERO	NOMBRE
2	Jesús Abad Guzmán López
4	María José Rodríguez Barrera & María Fernanda García Rodríguez
6	Alejandra Nicole Schafer Badillo
8	Marcos Ernesto Ramírez Ramírez

10	Salvador Alejandro Cuevas Villicaña
12	Cristopher Van Hout
14	Pablo E. Uribe Herrera
18	Ricardo Alonso Echevarría Solana
22	Rodrigo Aguilar Díaz
24	Valeria Gómez Vela
26	Manuel Rivera Cerón
28	Iván Eduardo Sedeño Jiménez
30	Alejandro Efraín Marín Perarlta
34	Sebastián Iturbe
36	Miriam Bravo López
38	Athena Tamayo Luisce
40	Victor Daniel Flores Ocampo

Abstracts de pláticas

Conferencia magistral

Nombre: Dr. Federico Sánchez Quinto
Jefe de grupo
Título: Paleogenómica de la Megafauna y el Paleoambiente de la Cuenca de México
Autores y afiliaciones:
Alejandra Castillo Carbajal 1, Eduardo Arrieta Donato 2, Vanssy Li 3, Miriam Bravo López 1, Ernesto Garfias Morales 1, Ángeles Taveres Guzmán 1, Rigoberto Padilla Bustos 1 , Viridiana Villa Islas 1, Alejandro López Jímenez 4, Mashaal Sohail 5, Camilo Chacón-Duque 3, Joaquín Arroyo-Cabrales 4, María C. Ávila Arcos 1
1) International Laboratory for Human Genome Research, Universidad Nacional Autónoma de México (UNAM), Querétaro, México.
2) Licenciatura en Ciencias Genómicas - UNAM - Cuernavaca
3) Centre for Paleogenetics, Stockholm, Sweden
4) Laboratorio de Arqueozoología, Subdirección de Laboratorios y Apoyo Académico, Instituto Nacional de Antropología e Historia, Mexico City, Mexico.
5) Centro de Ciencias Genómicas UNAM

Abstract:
Durante el Pleistoceno tardío el territorio contemporáneo de México albergó múltiples especies de megafauna extinta. La historia evolutiva de estas especies ha sido estudiada durante décadas con enfoques paleontológicos, arqueozoológicos y paleoecológicos. No obstante, a la fecha no se han aprovechado los avances del campo de la paleogenómica para un estudio genético de las mismas. Los recientes hallazgos paleontológicos recuperados durante la construcción del Aeropuerto Internacional Felipe Ängeles (AIFA) presentan una oportunidad única para investigar la historia evolutiva de estas especies en México y América. En esta plática se abordarán los resultados más recientes para la reconstrucción de la historia evolutiva de la megafauna extinta y el paleoambiente de la Cuenca de México. Los datos paleogenómicos de algunas de especies de megafauna analizadas implican un paradigma para el poblamiento de estos icónicos animales en América, enfatizando la importancia de realizar estudios de estas especies en México.

Regulatory Genomics and Bioinformatics Lab

Nombre: Sofia Guadalupe Salazar Magaña
Estudiante de licenciatura
Título: Studying the Systemic Lupus Erythematosus expression and contextualizing it in a Mexican cohort
Autores y afiliaciones:
Ana Laura Hernández-Ledesma(1), Evelia Lorena Coss-Navarrete (1), Karen Julia Nuñez-Reza (1), Diego Ramírez-Espinosa (1), Florencia Rosetti-Sciutto (2), Selene Lizbeth Fernández-Valverde (3), María Gutiérrez-Arcelus (4), Deshiré Alpízar-Rodríguez (5), Alejandra Medina-Rivera (1)
1: Laboratorio Internacional de Investigación sobre el Genoma Humano (LIIGH-UNAM)
2: Departamento de Inmunología y Reumatología, Instituto Nacional de Ciencias Médicas y Nutrición Salvador Zubirán (INCMNSZ).
3: University of New South Wales (UNSW).
4: Brigham and Women's Hospital, Harvard Medical School. Broad Institute. Boston Children's Hospital, Harvard Medical School.
5: Unidad de Investigación. Colegio Mexicano de Reumatología.

Abstract:
Systemic Lupus Erythematosus is an autoimmune disease where the immune system recognises and reacts against self-antigens, immune tolerance stops this process through mechanisms that are regulated by dendritic cells. In order to understand the genetic effects of this disease, we studied the immune response at a gene expression level, specifically using peripheral blood mononuclear cells (PBMC) which include important immune cells involved in the SLE etiology. We first conducted a differential expression analysis of bulk RNA-seq public data, with a total of 104 control and 214 SLE samples. We found an overexpression in SLE samples of genes associated with response to viruses and interferon signaling. We also analyzed data from the Mexican Lupus registry, which is a Mexican cohort that we created in order to improve diagnosis, treatment and follow up of SLE and it recovers demographic, clinical and biological (blood) data from SLE patients and controls. To get insights about immune tolerance as therapies in mexicans, from blood samples, monocytes were isolated and differentiated into monocyte-derived dendritic cells (moDCs) and monocyte derived tolerogenic dendritic cells (tolDCs), both of them also additionally supplemented with imiquimod (IMQ). We performed a differential expression analysis of all cell types between SLE samples and controls and found that patients have a distinct expression signature which is more notorious when the disease activity is higher.

Nombre: Evelia Lorena Coss-Navarrete
Posdoctorante
Título: Project JAGUAR:mapping immune cell diversity across Latin America
Autores y afiliaciones:
Olga Cuenca (1), Diego Ramírez-Espinosa (1), Alejandra Schäfer (1), Ana Hernandez-Ledesma (1), Marcela Sjoberg (2), Carolina Alvarez (2), Felipe Gajardo (2), Benilton de Sá Carvalho (3), Heitor Neto (4), Andreza Gama (4), Luis Tataje (5), Damaris Esquen (5), Maximiliano Berro (6), Marina Fernandez (7), Pablo Romagnoli (7), Danilo Ceshin (7), Yesid Cuesta-Arcos (8), Carlos Romero (8), Julieth Lopez (8), Jenny Gallo-Franco (8), Adriana Rojas (9), Liliana Lopez Kleine (10), Tarran Rupall (11), Thais de Oliveira (11), Matiss Ozols (11), Anna Lorenc (11), Carla Jones (11), Gosia Trynka (11), Alejandra Medina-Rivera (1).
1: Laboratorio Internacional de Investigación sobre el Genoma Humano (LIIGH-UNAM), Mexico.
2: Facultad de Ciencias Biológicas de la Pontificia Universidad Católica de Chile (PUC), Chile.
3: Universidade Estadual de Campinas, Brazil.
4: Faculdade de Farmácia da Universidade Federal do Rio de Janeiro (UFRJ), Brazil.
5: Facultad de Ciencias de la Salud - EPMH. Universidad Privada San Juan Bautista (UPSJB), Peru.
6: Hospital de Clínicas de la Facultad de Medicina, Universidad de la República, Uruguay
7: Centro de investigaciones en Medicina Traslacional ¨Severo R. Amuchástegui" (CIMETSA), Instituto Universitario de Ciencias Biomédicas de Córdoba (IUCBC), Argentina.
8: Instituto Colombiano de Medicina Tropical, Colombia.
9: Pontificia Universidad Javeriana - Bogotá, Colombia
10: Universidad Nacional de Colombia en Bogotá, Colombia.
11: Wellcome Sanger Institute, UK.
Abstract:
Project JAGUAR (Joining All: Genes, immUnity And diveRsity) is an international project in which will sequence immune cell diversity across Latin America for the first time, shedding light on how ancestry impacts the immune system and response to disease.
Latin American populations, including Mexico, comprise genetic and cultural admixture among European, Indigenous, African, and Asian groups, additionally shaped by the rich diversity of ecosystems and population migrations. This project seeks to define the diverse composition of peripheral blood mononuclear cells (PBMCs) in healthy individuals from seven distinct countries in Latin America. Our lab is responsible for sampling coordination and meta-data acquisition across LATAM.
This proposal uses a range of single-cell technologies (scATAC-seq, scRNA-seq, CITE-seq and whole genome sequencing) to identify how diverse ancestries impact gene expression and the composition of immune cells.
This research represents a remarkable opportunity to contribute to greater understanding of genetic and environmental influences on all Mexican traits.
In this presentation, I will focus on the advances we have had in all countries, the strategies we have employed, and invite all the public to leave a mark in history.

Cancer Genetics and Bioinformatics Lab

Nombre: Patricia Basurto Lozada
Estudiante de doctorado
Título: Genomic subtypes of acral lentiginous melanoma tumors associate with clinical outcomes
Autores y afiliaciones:

Martha E Vázquez-Cruz(LIIGH), Dennis Cerrato-Izaguirre (INCan), Christian Molina-Aguilar (LIIGH), Fernanda G Arriaga-González (Sanger Institute), Jesus Rene Chion Wong-Ramirez (LIIGH), Héctor Martínez-Said (INCan), Alethia Álvarez-Cano (INCan), Luis Alberto Tavares-de la Paz, Diego Hinojosa-Ugarte, Dorian Y García-Ortega (INCan), Marcos Díaz-Gay (UCSD), Ludmil B. Alexandrov (UCSD), Yesennia Sánchez-Pérez (INCan), Alfredo Hidalgo-Miranda (INMEGEN), Patricia A Possik (INCA), David J Adams (Sanger Institute), Carla D Robles-Espinoza (LIIGH).

Abstract:
Acral lentiginous melanoma (ALM) is the most frequent melanoma subtype in Mexico. Most of ALM's genomic knowledge comes from studies performed on European and Asian populations. To shed light into ALM genomics in the Mexican population we performed a genomic profiling of 128 ALM tumor samples from 96 Mexican patients. The genomic profiling was done through whole exome sequencing on tumor-nomal paired samples followed by somatic variant identification and identification of frequently mutated genes. We also performed estimation of copy number alterations (CNA) in 85 samples. From this study we identified NRAS, KIT, BRAF and NF1 as the most frequently mutated genes in the cohort. The regions frequently affected by CNA matched previous results from other populations, which included regions containing genes such as KIT, CDK4, CCND1 and CRKL. We then compared the burden of CNA between samples with different driver mutational status (NRAS, KIT, BRAF , NF1 and wild type), where we observed a lower burden of CNA in samples with mutations in BRAF and NRAS compared to samples with mutations in KIT. Samples harboring KIT mutations also seemed to have more whole genome duplications compared to samples with other mutations. Through correlation analysis with clinical data we also observed that patients with KIT mutations were associated with early recurrences. We hope that further investigation regarding KIT mutated tumors might help us find novel therapeutic options for ALM patients.

Nombre: Martha Estefania Vázquez Cruz
Estudiante de doctorado
Título: Transcriptional and spatial protein profiling of acral melanoma tumours from Mexican patients
Autores y afiliaciones:
Patricia Basurto-Lozada (LIIGH UNAM), Christian Molina-Aguilar (LIIGH UNAM), Ingrid Ferreira (Sanger Institute), Héctor Martínez-Said (INCan), Alethia Álvarez-Cano (Christus Muguerza Mty), Luis Alberto Tavares-de la Paz, Diego Hinojosa-Ugarte, Dorian Y. Garcia-Ortega (INCan), Julia M Martínez-Gómez (Universidad de Zurich), Mitchell P Levesque (Universidad de Zurich), Patricia A Possik (INCA Brasil), David Adams (Sanger Institute), Carla D. Robles-Espinoza (LIIGH UNAM; Sanger Institute).

Abstract:
Acral melanoma (AM), although overall a rare type of melanoma, is the most common form of the disease in a number of countries in Latin America, Africa, and Asia; it is associated with a poor prognosis and recurrence. In this study, we seek to gain a better understanding of the tumor-immune components of AM and their relationship to transcriptional programs. We performed transcriptome sequencing through exome-capture bulk RNA-seq on primary tumors from 64 Mexican patients and spatial protein profiling on tumor segments from 45 patients. Samples were collected at the National Cancer Institute of Mexico and have been annotated with vast clinical information. We identified differentially expressed genes such as CXCL8, MMP1, and TERT in ulcerated lesions. Deconvolution showed a high abundance of cancer-associated fibroblasts and the absence of NK cells. Consensus clustering identified three subgroups based on global gene expression. Additionally, we identified

differential protein abundance of fibronectin, VISTA, and B7-H3. A comparison between protein and RNA data for 35 targets is being conducted. AM is characterized by an immunosuppressive microenvironment, and our analyses point to genes that could drive important prognostic characteristics. The present study will enhance our understanding of the TME components and the antitumor response in an understudied disease.

Nombre: Felipe de Jesús Castañeda Córdova
Estudiante de maestría
Título: The transcriptomic landscape of the fibrotic liver under a time-restricted feeding regime
Autores y afiliaciones:
Arriaga González, Fernanda G (Sanger Institute); Robles Espinoza, C. Daniela (LIIGH); Simonin Wilmer, Irving (LIIGH); Rivera-Zavala, Julieta Berenice (ITESM); Molina Aguilar, Christian (LIIGH); Contreras, Kenya (LIIGH).

Abstract:
Chronic liver disease is a progressive condition with various stages (fibrosis, cirrhosis, and hepatocellular carcinoma) that can be replicated in animal models using diethyl-nitrosamine (DEN). Caloric restriction can delay the progression of liver damage. We aimed to characterize the effect of time-restricted feeding (TRF) on the induction of liver fibrosis in an experimental model. Forty male Wistar rats were randomly divided into four groups: AL (ad libitum), AL+DEN, TRF, and TRF+DEN. DEN treatment lasted for 8 weeks followed by a two-week clearance period. Groups under the TRF regime had food access for 2-hours every day. We collected biometric data, blood and liver samples, and visceral adipose tissue (VAT). We performed a complete blood count, blood chemistry, and histopathological analysis of liver tissue. Our transcriptomic analysis included differential expression analysis (DE), gene set enrichment analysis (GSEA), and gene interaction network analysis. We found that TRF led to a 40% decrease in food intake and a reduction in VAT mass. Histopathological and biochemical evaluation of liver biopsies and blood ruled out advanced stages of liver damage. Bioinformatics analysis identified unique changes in genes from various metabolic pathways for the TRF+DEN condition. Our theory is that caloric restriction improves the utilization of metabolic substrates, thus reducing the creation of reactive oxygen species and dampening the DEN's deleterious effect.

Ecology and Evolution Lab

Nombre: Dr. Sur Herrera Paredes
Jefe de grupo
Título: Comunidades sintéticas para diseccionar propiedades complejas
Autores y afiliaciones:
Sur Herrera Paredes1, Natalia Estrada Hernández1,3, Itzy Daniela Pérez Alvarado1,2, Natalia Said Muñoz1 & Carina Uribe Díaz1
1 Laboratorio Internacional de Investigación sobre el Genoma Humano,
2 Licenciatura en Ciencias Genómicas, Escuela Nacional de Estudios Superiores Juriquilla, Universidad Nacional Autónoma de México
3 Arkansas State University, Campus Querétaro

Abstract:
Actualmente, debido a su enorme potencial biotecnológico y relevancia ambiental, las comunidades bacterianas son el foco de intensos esfuerzos de caracterización. Sin embargo, la mayoría de las

funciones bacterianas de interés son propiedades complejas que resultan de interacciones entre la el contexto biótico y abiótico de un ecosistema. Estudios observacionales o que se enfocan en una sólo organismo están fundamentalmente limitados para diseccionar los mecanismos detrás de estas propiedades complejas. En principio, la ecología sintética nos permite diseccionar estas propiedades complejas, per el espacio de posibles comunidades es tan vasto, que ha resultado inviable explorar sistemáticamente la relación entre la composición de una comunidad y sus propiedades emergentes. En el laboratorio de ecología y evolución planteamos una estrategia para muestrear eficientemente el espacio de comunidades que nos permite inferir las contribuciones específicas de cada especie a las funciones de su comunidad. Nuestras simulaciones indican que esta estrategia mantiene el poder estadístico y estamos en proceso de construir las primeras comunidades en un gradiente de temperatura. Esperamos que este trabajo nos permita diseñar comunidades sintéticas con funciones predecibles y robustas ante las perturbaciones anticipadas por el cambio climático.

Paleogenomics and Evolutionary Biology Lab

Nombre: Alan Vladimir Godinez Plascencia
Estudiante de doctorado
Título: Evaluating in silico HLA typing of low-coverage, shotgun sequencing aDNA data
Autores y afiliaciones:
Federico Sanchez-Quinto - Paleogenomics and Evolutionary Biology, LIIGH-UNAM, México.
Mattias Jakobsson - Department of Organismal Biology, Uppsala University, Sweden.

Abstract:
Characterizing the HLA system from WGS data has proved crucial for various clinical and research applications in immunology, and a constant increase in broadly accessible data has made it possible for computational strategies to surge as an effective method for doing so. Moreover, recent developments in capture techniques of aDNA data have allowed researchers to take a glimpse into the immunogenic makeup of individuals from the past, uncovering questions regarding the evolution of this system in specific historical contexts. However, it remains to be seen whether these methods are suitable for shotgun-sequencing aDNA data. To investigate this, we evaluated the performance of in silico HLA typing for Class I genes at the single and second-field resolution, assessing the accuracy achieved on WGS data with simulated aDNA profiles as well as the concordance displayed on ancient genomes at different levels of read depth. This evaluation allowed us to construct a framework for the characterization of HLA alleles at different levels of genomic coverage, showing the feasibility and areas of opportunity for in silico HLA typing using shotgun aDNA data. We used this framework to explore the immunological landscape throughout time in Europe, where we found marked shifts in the frequencies of Class I HLA alleles associated with disease from prehistoric times. Furthermore, we started analyzing HLA evolution through the colonization of America using the insight provided by these analyses.

Nombre: Eduardo Alejandro Arrieta Donato
Estudiante de licenciatura
Título: Evaluación de imputación en indígenas mexicanos usando paneles de referencia con diferente ancestría
Autores y afiliaciones:
Federico Sánchez Quinto

International Laboratory for Human Genome Research, Universidad Nacional Autónoma de México (UNAM), Querétaro, México.

Abstract:
Estudios genómicos requieren gran cantidad de muestras para dar solidez estadística a sus inferencias. Por financiamiento limitado, los laboratorios prefieren abarcar la mayor cantidad de muestras usando arreglos de genotipificación o secuenciación de baja cobertura, los cuales usualmente no capturan toda la información genética de las muestras. Para maximizar la información genética disponible es posible usar métodos computacionales para imputar las variantes alélicas no observadas provenientes de un panel de referencia de imputación. Actualmente, los paneles de referencia de imputación existentes tienen subrepresentadas ancestrías indígenas de América y México, lo cual puede afectar negativamente a la precisión de imputación en individuos con dicha ancestría. En este trabajo se imputaron individuos indígenas mexicanos sub-muestreados a bajas profundidades de cobertura, para comparar los alelos imputados contra el llamado de variantes original. La imputación se realizó utilizando distintas composiciones de ancestría para los paneles de referencia. Los resultados sugieren que la precisión de imputación de sitios polimórficos "privados" para México depende fuertemente de usar paneles de referencia que contengan la ancestría indígena regional correspondiente a las muestras evaluadas; particularmente para sitios de baja frecuencia alélica en la población mexicana.

Nombre: Leonardo Yair Correa Mendoza
Estudiante de licenciatura
Título: Demographic origin of individuals found in the Sa Galera sanctuary (Spain) from paleogenomic data
Autores y afiliaciones:
José Jorge Argüello Menéndez2, Ramón MartínGordon2, Anders Götherström2, Cristina Valdiosera3, Federico Andrés Sánchez Quinto4
1 Center of Genomic Sciences, National Autonomous University of Mexico, Cuernavaca, Morelos, Mexico.
2 Department of Archaeology and Classical Studies, Stockholm University, Stockholm, Sweden
3 University of Burgos, Department of History, Geography and Communications, Burgos, Spain
4 International Human Genome Research Laboratory, National Autonomous University of Mexico, Juriquilla, Querétaro
Abstract:
The study of ancient DNA opens the doors to a better understanding of events and evolutionary processes in our past. There are currently multiple paleogenomic studies that use genetic data from samples throughout the European continent to investigate the dynamics of demographic events and the social structure of populations over different periods. This is the case of the sanctuary of Sa Galera, which is located on an islet in the Bay of Palma in Spain. The islet has a history of being inhabited by Phoenicians, as evidenced by the presence of ceramic remains. We found 8 individuals dated between 200 cal BCE and 100 cal CE, indicating dwelling during the end of the Bronze Age. In this project, we investigate the demographic origin of the individuals of Sa Galera and whether the individuals who lived during the same period may have been related to each other. The genetic affinities of the exhume individuals suggest that they had different demographic origins. This result implies that people from various cultural backgrounds lived together on the same island during the same period. This observation aligns with recent paleogenomic studies indicating high population mobility in Bronze Age Europe from various geographic regions. The results provide more information about the demographic

dynamics of migrations in the Mediterranean during the Bronze Age, in particular of individuals associated with the Phoenician culture according to the remains found associated with them.

Evolutionary Systems Biology Lab

Nombre: Emmanuel Hernández Sánchez
Estudiante de licenciatura
Título: Dinámica evolutiva de un modelo del reloj circadiano de Neurospora crassa
Autores y afiliaciones:
Mariana Gómez-Schiavon, LIIGH e iBio.

Abstract:
Neurospora crassa es un hongo y organismo modelo, el cuál ha sido muy estudiado en cuanto a su reloj circadiano. Se han propuesto ya muchos modelos matemáticos para estudiar las propiedades de este reloj circadiano. Los modelos van desde aquellos muy simples enfocados en estudiar las oscilaciones del reloj circadiano, y aquellos más complejos que estudian su capacidad para compensar las fluctuaciones ambientales, por ejemplo cambios de temperatura o de glucosa. Sin embargo, las propiedades de las oscilaciones del reloj circadiano no se han estudiado en profundidad. En el mismo sentido, existen pocos modelos que consideren la presencia y el efecto del ruido bioquímico, y ninguno que sepamos ha explorado la dinámica evolutiva del circuito de regulación génica del reloj circadiano. En este proyecto pretendemos evaluar la robustez de las oscilaciones en un modelo de circuito de regulación del reloj circadiano de Neurospora crassa. Esto contribuirá a nuestra comprensión de su relevancia en la adaptación y anticipación a un entorno fluctuante, y las propiedades específicas responsables.

Nombre: Manuel Eduardo Hernández García
Estancia académica
Título: Cuantificando el impacto de las fluctuaciones extrínsecas en el sistema p53.
Autores y afiliaciones:
Mariana Gómez Schiavon(LIIGH-UNAM)

Abstract:
En este trabajo, se estudia el efecto de las fluctuaciones extrínsecas en sistemas biológicos, brindando una visión más realista de los montajes experimentales y los sistemas biológicos. Presentamos un enfoque analítico alternativo para modelar el sistema mediante un conjunto de ecuaciones diferenciales ordinarias. El estudio se enfoca en el sistema p53, vital en la respuesta al estrés celular, la regulación del ciclo celular y la inducción de apoptosis. Nuestro objetivo fue cuantificar cómo las oscilaciones del sistema se ven afectadas por las fluctuaciones extrínsecas, específicamente las relacionadas con la temperatura.

Genome Evolution Lab

Nombre: Samuel Quiñones Galeana
Estudiante de licenciatura
Título: Exploring Agave Yeast Biodiversity for Bioethanol Production
Autores y afiliaciones:

Keertna Bhuvan-University of Illinois at Urbana Champaign. Christopher, V Rao-University of Illinois at Urbana Champaign. Lucia Morales Reyes- Laboratorio internacional de investigación sobre el genoma humano.

Abstract:
The imminent global crisis caused by global warming due in part to the amassing of greenhouse gases requires quick and effective solutions. The integration of microorganisms for developing alternative energy sources with equivalent power capacity but lower pollution levels is a valuable and viable option. One option is bioethanol, notable for having a similar octane number to gasoline. Bioethanol is produced from the fermentation of lignocellulosic biomass (LCB). The latter shares characteristics with agave juice, from which a collection of genetically diverse yeasts has been isolated. The project's objective is to evaluate the growth of these agave yeasts on LCB as well as their bioethanol production. To achieve this, 24 yeasts isolates were selected, including 18 S. cerevisiae, 3 S. paradoxus, and 3 hybrids of between these two species, encompassing the wide genetic diversity found in agave fermentations. All selected isolates were able to grow in 100% sorghum hydrolysate (SH), an achievement that a laboratory strain failed after 96 hours of incubation. Additionally, the two best S. cerevisiae growers produced around 18g/L of ethanol in 24 hours in 50% SH, which is competitively comparable with other strains in the literature. In conclusion, the agave yeasts strains here studied have potential for bioethanol production using SH. Furthermore, their ethanol production and adaptation to SH can be enhanced through genetic editing techniques, making them even more competitive.

Nombre: Mariana Guadalupe Guerrero Osornio
Estudiante de doctorado
Título: Time-lapse of the microbial composition during agave fermentation using Meta-HiC
Autores y afiliaciones:
Dra. Lucía Morales Reyes

Abstract:
The fermentation step in the production of traditional Mexican distilled spirits stands out as it occurs in open tanks, promoting microorganism exchange with the surroundings. This fosters a niche with vast, yet uncharacterized, microbiological diversity. Variations in microbial communities throughout agave juice fermentation have been established. Recent unpublished studies identified yeast hybrids and their parental species Saccharomyces paradoxus and Saccharomyces cerevisiae within tanks. The research aims to describe microbial communities at the species level during agave fermentation, utilizing Meta-HiC for sequencing, assembling, and annotating MAGs. Hybrids analyses will determine their relative abundance along the fermentation phases in which hybrids and parental species coexist. In this study, I present the proportion of reads assigned by Kraken2, these reveal that 10%-25% of the reads remain unclassified (Database: PlusPFP). At the end of the fermentation 98% of the reads were classified for bacteria, revealing a high abundance of this domain. We uncovered the potential presence of viruses and archaea, which had not been previously reported in such environments. So far, the Meta-HiC analysis yielded 141 bacterial genome bins from the metagenomic data of all the fermentative phases. Parental species presence was confirmed by tracking their genome coverage. Further analysis is needed to gain insights into the succession and characterization of this fermentation.

Population and Evolutionary Genomics Lab

Nombre: Laura Carrillo Olivas

Estudiante de doctorado

Título: Exploring Ancient Pathogens in Colonial Mexico City through Paleogenomics

Autores y afiliaciones:

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Abstract:

Following European conquest, epidemics devastated Indigenous populations in the Americas. These outbreaks were linked to the introduction of pathogens through colonization and the trade of African Enslaved individuals. While diseases like measles was common, others like the Cocoliztli with unknown causal agents also emerged. To capture the diversity of pathogens in the colonial Mexican population, molecular tools are necessary for specific identification. We used the paleogenomics toolkit to screen 97 individuals dated to the colonial period in Mexico City in search of ancient pathogen DNA. These were chosen from El Templo de la Inmaculada Concepción 'La Conchita' (n=15) and Hospital Real San José de los Naturales (n=82). Sequencing data was compared with the human genome. While the non-human fraction of reads was taxonomically classified using Kraken2, and selecting pathogenic species through a complementary approach that identified coincidences with the virulence factors database VFDB2022. After applying standard aDNA quality filtering criteria, Streptococcus pneumoniae was identified at both sites. In HSJN, a variety of pathogens were found, including Tannerella forsythia, Salmonella enterica Paratyphi C, and Mycobacterium tuberculosis. These results offer insight into health and infectious diseases in that period, inciting further phylogenetic studies, to identify potential epidemiological distinctions between these ancient strains and those in the present population.

Nombre: Miguel Alejandro Navarro

Estudiante de licenciatura

Título: Paleoproteomics and Ancient Pathogen Identification

Autores y afiliaciones:

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6. National Institute of Anthropology and History, CDMX, Mexico
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Abstract:
Proteins are a rich source of evolutionary information, capable of withstanding degradation better than nucleic acids. However, to our knowledge, the use of paleoproteomics to identify and study ancient pathogens remains very limited, making it an exciting new niche worth exploring.
Thanks to the collaboration of the National Institute of History and Anthropology in Mexico, we had access to 6 samples from individuals dating back to the 17th century who were previously reported positive for ancient DNA of different human pathogens. Taking a shogun proteomics approach the extracted peptides were injected in an LTQ Orbitrap Velos, and resulted files raw files were processed using MaxQuant v.2.0.3.0 for peptide detection.
Besides the various peptides from environmental bacteria, we detected numerous peptides of pathogenic bacteria from the genus Salmonella, as well as several peptides from Tanerella forsythia, a widely studied oral pathogen. Furthermore, the analysis showed the presence of three different human viruses, Hepatitis B, Herpes Simplex 1, and Rabies.
We expect this exploratory study to highlight the potential of paleoproteomics in pathogenic research and the utility of a multidisciplinary approach, especially when working to unravel the mysteries of ancient epidemics.
Keywords: Paleoproteomics, Paleogenomics, pathogens, epidemics.

Mendelian Genomics and Precision Health Lab

Nombre: Tania Sepulveda Morales
Estudiante de licenciatura
Título: Estudio de percepciones de la población general en México acerca de las enfermedades raras
Autores y afiliaciones:
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Abstract:
Las enfermedades raras (ERs), son aquellas que tienen una baja prevalencia en la población general. En conjunto, estas afectan a alrededor del 8% de la población mundial. A pesar de su impacto en la sociedad, hay escasa información disponible.
Se elaboró un cuestionario en línea de 33 preguntas para obtener información del conocimiento de la población mexicana sobre ERs y conceptos asociados de genética. Este se lanzó el 27 de octubre de 2023 a través de las redes sociales de la Red Mexicana de Enfermedades Raras (ReMexER). Al 30 de enero de 2024, 224 personas habían participado; el 87.1% tenía conocimiento de las ERs y el 51.8% conoce a alguien con una enfermedad rara. La mayoría de los encuestados percibe a las ERs como padecimientos de baja prevalencia, difícil diagnóstico y de causa desconocida, afectando principalmente a niños menores de 5 años. La gran mayoría de los participantes reconoce la genética

como la causa principal de ERs, pero sólo el 49.1% sabe que pocas ERs tienen un tratamiento disponible.
En general, los participantes tienen una percepción acertada acerca de las ERs; sin embargo, gran parte de ellos son miembros de la comunidad extendida de ReMexER, lo que sugiere una sensibilización previa respecto a las ERs. Esfuerzos adicionales son necesarios para diversificar la muestra a fin de obtener datos más representativos de la población general en México.

Nombre: César Enrique Calvo Aspiros
Estudiante de licenciatura
Título: Perspectivas sobre el Diagnóstico: Registro Mexicano de Enfermedades Raras
Autores y afiliaciones:
Claudia Gonzaga Jáuregui - Laboratorio Internacional de Investigación sobre el Genoma Humano, Universidad Nacional Autónoma de México, UNAM Campus Juriquilla, Querétaro, México el Diagnóstico: Registro Mexicano de Enfermedades Raras.

Abstract:
En el contexto de la salud en México, las enfermedades raras y poco frecuentes representan un desafío significativo en términos de diagnóstico, tratamiento y atención médica que afecta a aproximadamente 10 millones de personas en todo el país. En 2022 se inició el "Estudio de Prevalencia y Necesidades de Diagnóstico de Enfermedades Raras y Poco Frecuentes en México" con el objetivo de conocer más sobre estas condiciones y quienes viven con ellas en México a través de un cuestionario electrónico abierto al público. Los resultados del primer año del Registro se basan en las respuestas de 144 pacientes mexicanos afectados por enfermedades raras y poco frecuentes. La falta y retraso en la obtención de un diagnóstico preciso es un reto significativo para estos pacientes, con impactos negativos en su calidad de vida y el manejo de su condición. El diagnóstico molecular ofrece una herramienta poderosa para identificar las causas genéticas subyacentes de estas enfermedades, sin embargo, el acceso a pruebas genéticas sigue siendo limitado debido a barreras financieras y de infraestructura en México. El impacto económico y emocional durante la odisea diagnóstica de los pacientes que viven con enfermedades raras representa un desafío importante para ellos y sus familias.

Nombre: Issis Abril Pérez Alvarado
Estudiante de maestría
Título: Identification of Genomic Variants Associated with Hereditary Cancer in the Mexican Population
Autores y afiliaciones:
Tania Sepulveda-Morales, Cristopher Van Hout, Claudia Gonzaga-Jauregui. International Laboratory for Human Genome Research, Laboratorio Internacional de Investigación sobre el Genoma Humano, Universidad Nacional Autónoma de México, Juriquilla, Querétaro, México.

Abstract:
Hereditary cancer contributes to a significant proportion of total cancer morbidity. Breast, colorectal, and prostate cancers hold the highest prevalence and mortality rates. Population genomic studies facilitate the identification of genetic risk variants to estimate disease prevalence in a population. However, genomic data from European ancestry individuals comprise >90% of genomic data available, whereas <0.5% represent Latin American individuals.
We analyzed data from the Mexico City Prospective Study (MCPS) to identify genomic variation in 90 genes associated with hereditary cancer. We identified 864 variants present in 72 genes, 421 were novel loss-of-function variants and 443 were previously reported variants. The genes with the greatest number

of variants were LZTR1, FANCA, ATM, FANCM and RAD50, representing 34.2% of the total identified variants. In addition, the genes with the highest number of carriers were RAD50, FANCA, LZTR1, RNASEL and NBN. Altogether, 4.2% of the individuals in the MCPS cohort carry a relevant hereditary cancer variant.
Our results suggest the existence of relevant risk, and potentially medically actionable, variants segregating in the Mexican population that are associated with susceptibility to a wide range of cancers (mainly breast, colorectal, and prostate cancer) consistent with the epidemiological data. Carriers of these variants could benefit from early screening and timely medical interventions that may reduce morbidity and mortality.

Statistical Genomics and Population Health Lab

Nombre: Leonardo Hernandez Delgado
Estudiante de licenciatura
Título: Unraveling the genetic basis of Keloids in African Yoruba Individuals
Autores y afiliaciones:
Cristopher Van Hout & Claudia Gonzaga-Jauregui
International Laboratory for Human Genome Research, UNAM, Queretaro, Mexico

Abstract:
The formation of a scar can be thought of as a complex mechanism that has evolved over millions of years to restore functionality, not an esthetic aspect. This process is susceptible to abnormalities that may result in an aberrant scar, this is known as keloid. Keloids are often accompanied by pain and social stigma for the notorious scar. Keloid formation mechanisms are poorly understood, and current treatments are ineffective. Different studies have evidenced the genetic component of Keloids, including both familiar studies and previous GWAS on Asian and European ancestries. To date, no genome-wide association studies have been conducted in the African population. To investigate the genetic basis underlying the development of keloid, the Yoruba African group presents an opportunity for this goal since Keloids prevalence in their population has been documented for centuries. Interest in conducting genetic research with this ethnic group led to a recruitment study in Nigeria led by researchers from the University of Connecticut, along with Nigerian Researchers. 4200 individuals provided phenotypic information and consent to obtain blood or saliva samples for further genetic studies. We applied a Genome-Wide Association Study (GWAS) on keloid phenotype.
Signals obtained from the Association study replicate NEDD4 (a previously associated gene with keloids from previous GWAS). Further research in the results obtained led to interesting discoveries in Keloids Research.

Nombre: Dr. Cris Van Hout
Jefe de grupo
Título: Ongoing projects and early ideas in the Statistical Genomics and Population Health group
Autores y afiliaciones:

Abstract:
Themes of ongoing work and early ideas from the Statistical Genomics and Population Health research group at the LIIGH are presented. Various projects including Amish/Mennonite population structure, Nigerian exome sequencing, and alternative models for genotype-phenotype association are motivated.

Major research focus in the lab now includes the Mexico City Prospective Study (MCPS), the largest genetic dataset in the world with ascertainment of individuals of Latin American ancestry. The MCPS contains exome data for 150,000 participants, metabolomics data, questionnaire data on risk factors and common disease. Prior analyses show high obesity and diabetes rates, which the lab has plans to refine and extend in subsequent projects. In some cases, it may also be useful to leverage the UK Biobank project to which the lab now has approved access to extend or replicate relevant results. Analyses for these big data genomics datasets are enabled in state of the art cloud compute environments. We argue that complex molecular models of disease require non-additive analyses with emphasis determined by public health impact.

Computational Population Genetics

Nombre: Valeria Alejandra Añorve Garibay
Estancia académica
Título: The contribution of archaic introgression to the heritability of complex traits in Mexico
Autores y afiliaciones:
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Center for Computational Molecular Biology, Brown University, Providence, RI 02912

Abstract:
The impact of archaic introgressed mutations to the genetics of complex traits and diseases has been recently studied in European populations. This line of research has found that archaic introgression is an important phenomena shaping the evolution of traits, such as the resistance to different pathogens or the metabolization of drugs. However, there has not been any research conducted on the contribution of archaic introgression to complex trait variation in Admixed populations from the Americas. To address this gap, we apply a reference-free method for detecting archaic introgression on the Mexican Biobank Project (MXB) data and quantify the contribution of archaic introgressed variants to metabolic and anthropometric traits from MXB. We infer a map of putatively Neanderthal and Denisovan archaic introgressed segments identified in Mexican individuals. We examine the contribution of these introgressed alleles to the heritability of a range of complex traits and diseases. This study provides the first rigorous basis for understanding the impact of archaic introgression on complex trait variation in Mexico and represents a valuable resource for future studies to gain power at identifying archaic introgression in underrepresented populations. Our analysis will be helpful for future studies to characterize the impact of archaic introgressed variants on complex traits.

Nombre: Héctor Alessandro López Hernández
Estudiante de doctorado
Título: Genética de Poblaciones Espacio-Temporal
Autores y afiliaciones:
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Laboratorio Internacional de Investigación sobre el Genoma Humano, Universidad Nacional Autónoma de México; Querétaro, México

Abstract:
Con el avance de las tecnologías de secuenciación estamos obteniendo una cantidad cada vez mayor de datos genómicos de centenas de miles de individuos procedentes de diversas partes del mundo y de distintas épocas abriendo la posibilidad de comprender mejor el impacto de distintos fenómenos evolutivos. El análisis de datos genómicos procedentes de distintas temporalidades y coordenadas geográficas es de particular interés para conocer la influencia de la selección natural en el genoma de los seres vivos a lo largo del tiempo y el espacio. En este trabajo presento una nueva metodología basado en un enfoque de máxima verosimilitud para inferir la tasa de dispersión geográfica de alelos y el impacto de la selección natural actuando en nuevas variantes genéticas mediante el análisis de las variaciones en las frecuencias alélicas de distintas poblaciones en diferentes momentos temporales. Los datos provienen de un grupo de personas del continente europeo además de partes de Asia, abarcando desde los 10000 años en el pasado hasta el presente. Con la información disponible de la literatura, analizo los alelos de interés médico más relevantes e infiero el impacto del flujo genético y la selección natural en los cambios en frecuencia de esos alelos a lo largo del tiempo. Los resultados de mis inferencias muestran una tasa de dispersión alta, que los alelos putativamente deletéreos son correctamente inferidos como deletéreos y que algunos alelos ventajosos varían en su valor.

Nombre: Alan Raymundo Izarraras Gomez
Estudiante de doctorado
Título: Inferring the impact of natural selection across the genome using local genealogies
Autores y afiliaciones:
Diego Ortega Del Vecchyo (LIIGH - UNAM)

Abstract:
Genetic variation can be classified in terms of its effect on fitness, expressed as selection coefficients (s). The distribution of fitness effects (DFE) measures the probability of a new mutation having a certain value of s, and as such, is central to understanding fundamental questions in population genetics. The DFE has previously been inferred in humans and other species using population-wide summary statistics of nucleotide sequence data. A drawback of these methods is the focus on specific summary statistics of the data that might not reflect the fine scale impact of natural selection. Here, we develop a new methodology to estimate the DFE using the collection of local genealogies that are embedded in the Ancestral Recombination Graph (ARG). The ARG contains the full history of coalescent and recombination events on the genomic sequence analyzed. We show how we can leverage the information contained on the ARG to estimate the impact of natural selection with more accuracy than previous methods that use summary statistics. We use a Monte Carlo approach in order to build a Maximum Likelihood (ML) framework using information from the number of lineages with a derived allele at discrete time points. Furthermore, we show that we can integrate the information from multiple trees to obtain estimates of the DFE. With simulated data, we demonstrate that inferences of discrete selection coefficients and parameters of the DFE are robust.

Abstracts de pósteres

Regulatory Genomics and Bioinformatics Lab

Póster #6

Alejandra Nicole Schafer Juarez Badillo
Estudiante de doctorado

Título: Mapping the diversity of immune cells in the Mexican population without disease.

Autores y afiliaciones:
Coss-Navarrete Evelia Lorena 1, Cuenca-Micó Olga 1, Hernandez-Ledesma Ana Laura 1, Ramírez Diego 1, Jones Carla 2, Lorenc Anna 2, Rupall Tarran 2,, Trynka Gosia 2, Medina-Rivera Alejandra 1.
1 Laboratorio Internacional de Investigación sobre el Genoma Humano, UNAM. Querétaro, México.
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Abstract
The immune system protects our organisms from pathogens. Various cells play a role in this defence, including those found in peripheral blood mononuclear cells (PBMCs) circulating in the blood. Differences in immune system responses exist among populations due to genetic variations, which affect disease prevalence. Understanding these genetic differences between populations is crucial. However, most of the functional genomic studies are done in European populations, lacking the presence of people living in latin american. In this project, we aim to analyze PBMCs from healthy Mexican donors, analyzing genetic information in these immune cells under specific stimuli. This will help us find genetic variants relevant to the Mexican population and enable us to study the expression of quantitative trait loci (eQTLs). This research will significantly impact the identification of relevant genetic mechanisms involved in the development of infectious and autoimmune diseases and their severity in the Mexican population, which could set the basis for developing therapeutic strategies in the future.

Póster #9

Ian Manuel Espinosa Méndez
Estudiante de licenciatura

Título: Heritability & Genetic Correlation of Cortical Surface Area & Thickness in Mexican Population

Autores y afiliaciones:
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Abstract
Cortical volume, cortical surface area, and cortical thickness are commonly studied as brain morphology traits. However, these measurements are often used interchangeably, despite cortical volume being the product of cortical surface area and cortical thickness, and the latter two appear to show no genetic

correspondence. This is problematic as many studies on aging and neuropsychiatric disorders have an incomplete picture of the architecture of the cerebral cortex by only assessing cortical volume or thickness. Additionally, most studies have examined the heritability and genetic correlation of these traits on populations of European and Asian ancestry, and less research on this matter has been conducted on the genetically admixed, Latin American populations. In this study we aim to characterize the heritability and genetic relationship between cortical surface area and cortical mean thickness, both in the overall cortical structure and in specific lobes/hemispheres in a sample of Mexican twins (63 monozygotic pairs and 32 dizygotic pairs) from the Mexican twin registry, TwinsMX. The heritability for the cortical surface area and mean cortical thickness were 68.89% and 27.04%, respectively. On the other hand, the cross-trait genetic correlation between traits was rA=0.34 and they had a phenotypic correlation rP=0.16. In contrast to the previous studies, we found a genetic overlap between both phenotypes which is more in line with other whole genome studies.

Póster *#17*

Luis Miguel Pedraza Meza
Estudiante de licenciatura

Título: Clinical, psychosocial and demographic factors affect decisions in SLE people.

Autores y afiliaciones:
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2 Escuela Nacional de Estudios Superiores Juriquilla.

Abstract
Como variables predictoras, se registraron la dosis diaria consumida de glucocorticoides, además de otros datos demográficos, psicosociales y clínicos. Las variables a predecir fueron dos; las elecciones temporales y las elecciones sociales, las cuales se evaluaron mediante dos paradigmas experimentales estándar (paradigma de la recompensa diferida y paradigma de los bienes comunes, respectivamente). Los resultados en los modelos de regresión, muestran un efecto entre el consumo de glucocorticoides y las decisiones temporales y sociales. Mayor dosis de glucocorticoides se asocia con tendencia a decisiones sociales vengativas, así como con la preferencia a recompensas inmediatas. Estos resultados sugieren que, el consumo de glucocorticoides en dosis altas tiene efectos en la toma de decisiones de personas con lupus. Por un lado, la preferencia por las recompensas inmediatas, en lugar de mejores recompensas futuras, podría asociarse con un deficiente apego al tratamiento médico. Y, por el otro lado, las reacciones severas ante interacciones sociales adversas, podrían asociarse con mayores índices de ira, hostilidad, ansiedad y depresión.

Póster #40

Victor Daniel Flores Ocampo
Estudiante de licenciatura

Título: Evaluating Parkinson's disease Polygenic Risk Score (PRS) transferability from European cohorts to latinx populations.

Autores y afiliaciones:
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2. Laboratorio Internacional de Investigación sobre el Genoma Humano.
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4. Instituto de Neurobiología UNAM.
5. Facultad de Psicología UNAM.
6. Global Parkinson's Genetics Program

Abstract
This study aims to develop a Polygenic Risk Score (PRS) for Parkinson's Disease within a diverse GP2 cohort. The predictive performance of state-of-the-art tools is assessed, with a focus on comparing ancestry-aware methodologies to baseline techniques. Emphasizing practical considerations related to data constraints, we explore scenarios using only European GWAS data, as it is the most common scenario for underrepresented background studies . Additionally, we assess the potential benefits of employing ancestry-specific GWAS (LargePD) for our samples with appropriate tools, contributing to the understanding of PRS application in underrepresented populations.

Cancer Genetics & Bioinformatics Lab

Póster *#10*
Salvador Alejandro Cuevas Villicaña
Estudiante de licenciatura
Título: Evaluación experimental del efecto oncogénico del gen ESR1 Y537C en células de melanoma A375
Autores y afiliaciones:
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1 Laboratorio Internacional de Investigación sobre el Genoma Humano, Universidad Nacional Autónoma de México, Campus Juriquilla, Querétaro.

Abstract
En un estudio realizado por Robles-Espinoza y colaboradores, a partir de los datos de secuenciación de genoma completo de 314 individuos de 135 familias del Reino Unido, se realizó un análisis bioinformático de identificación de variantes genéticas presentes en las familias con melanoma. Se encontraron variantes en 13 que podrían estar asociadas al melanoma, estos genes fueron: REST, ESR1, ENDOG, KRT18, CR, CALCA, KRT18, S100A7A, AGXT, WRN, MEPS2, KRT18 y KRT16. Después de una búsqueda bibliográfica y priorización de los efectos de las diferentes variantes de los 13 genes se encontró que la variante Y537C del ESR1 es la que tiene mayores antecedentes de estar asociada al desarrollo de cáncer, principalmente en mama.
El gen ESR1 codifica para el receptor a estrógenos alfa (REα) y ya ha sido ampliamente estudiado en cáncer de mama, en donde la sobreexpresión de este gen se ha asociado a pacientes con peor diagnóstico y pronóstico de la enfermedad. En el laboratorio de Genética de Cáncer y de bioinformática, perteneciente al Laboratorio Internacional sobre el Genoma Humano (LIIGH) se ha estado trabajando en los últimos años en demostrar que el posible efecto oncogénico de la mutación Y537C del gen ESR1 en la línea celular de fibroblastos de ratón NIH 3T3. El presente trabajo busca caracterizar el posible efecto oncogénico que el ESR1 Y537C tiene en la inducción de melanoma en la línea celular de melanoma A-375.

Póster #30
Alejandro Efraín Marín Peralta
Estudiante de licenciatura

Título: Local Ancestry-based GWAS of Acral Lentiginous Melanoma

Autores y afiliaciones:
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Abstract
Acral lentiginous melanoma (ALM) is an uncommon skin cancer with unknown etiology, albeit genetic risk factors have been proposed. Thus, in Mexicans, an admixed population, most of melanoma diagnoses are ALM. In the other hand, Genome-Wide Association Studies of admixed populations can boost their power by taking into account local ancestry. Here, we hypothesize that there are loci associated with a greater risk of developing ALM in the Mexican population, which can be uncovered by considering local ancestry.

Póster #35

Kenya Lizbeth Contreras Ramirez
Estudiante de doctorado

Título: Estudio del genoma completo del melanoma lentoginoso acral en pacientes mexicanos

Autores y afiliaciones:
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Abstract
El melanoma es una neoplasia maligna de los melanocitos considerado como el más agresivo dentro de los cánceres de piel ya que representa sólo el 4% de los tumores dermatológicos pero causa el 80% de las muertes. Este se clasifica en dos tipos clínicos: las lesiones cutáneas, y no cutáneas (acral, uveal y mucoso). El melanoma lentiginoso acral (MLA), en el segundo grupo, es una variante relativamente poco frecuente, que se caracteriza por afectar las plantas de los pies, las palmas de las manos o el lecho subungueal . La incidencia de este subtipo es análoga entre poblaciones, sin embargo, se considera como el más común para las poblaciones asiáticas, africanas y latinas, incluyendo a México. A la fecha la etiología del MLA es aún desconocida y presenta dificultades en cuanto a su tratamiento y diagnóstico. En estudios previos en pacientes de ascendencia europea se han encontrado diferencias en su perfil genómico respecto a otros melanomas. En este proyecto, pretendemos utilizar datos de secuenciación de genoma completo de pacientes mexicanos, no sólo para descubrir genes, mutaciones, variantes estructurales y firmas mutacionales asociadas con el riesgo y el desarrollo del MLA, sino apoyar la integración y el soporte con otras técnicas y estudios que eventualmente permitan develar los procesos mutacionales operativos en este tipo de cáncer y ampliar las opciones terapéuticas dirigidas a los pacientes, desde el estudio de un perfil poblacional poco estudiado pero afectado.

Póster #19

María Fernanda Talavera Cruz
Estudiante de licenciatura
Título: Mutational landscape of lung tumours from mice exposed to E-cigarette
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Abstract
E-cigarette usage has surged in recent years, raising concerns about its potential carcinogenicity. To elucidate the molecular underpinnings of e-cigarette-induced lung tumorigenesis, we conducted whole exome sequencing (WES) of lung tumors from mice exposed to e-cigarette smoke. Our analysis identified KRAS and BRAF as the most significantly mutated genes, with predominantly activating mutations. These findings underscore the oncogenic potential of e-cigarette exposure and provide insights into the molecular mechanisms driving lung tumorigenesis in response to e-cigarette use.

Póster #27
Christian Molina Aguilar
Técnico académico
Título: Efecto anti-inflamatorio de la restricción calórica en un modelo de fibrosis en hígado de rata
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Abstract
La fibrosis hepática (FH) representa un estadio temprano de enfermedades que puede evolucionar a cirrosis y a cáncer hepatocelular (HCC). La cirrosis induce insuficiencia hepática y es una de las principales causas de muerte (110 causa de muerte). El HCC es la 40 causa de muerte a nivel mundial por cáncer. Dadas estas cifras y aunado a que la fibrosis es una enfermedad reversible, existe un gran interés por conocer a fondo los mecanismos que inducen fibrosis y así contrarrestar los problemas relacionados a la cirrosis y HCC.
La FH es causada por estados de inflamación crónica que exacerban el estrés oxidativo y de la producción de fibras de colágena de la matriz extracelular. Mediante un modelo de inducción de FH en ratas macho Wistar, con el uso del hepatotóxico Dietilnitrosamina (DEN), hemos puesto a prueba el efecto protector de la restricción calórica (RC) que reportamos en cirrosis y HCC en 2017 (DOI: 10.1093/carcin/bgx052) pero ahora en estadio fibrótico.
Nuestros resultados preliminares sugieren: A) Las ratas con DEN exhibieron FH confirmada con depósitos de colágeno leves. B) Las ratas con RC redujeron un 30% su ingesta de alimento sin presentar malnutrición. C) Sin embargo, el análisis de RNAseq sugiere que la RC induce una disminución tanto

en las vías inflamatorias agudas como crónicas. E) Niveles de TNF-alfa confirman reducción de inflamación.
Hasta ahora nuestros datos sugieren que la RC genera un efecto anti-inflamatorio a nivel celular.

Ecology and Evolution Lab

Póster #26
Manuel Rivera Cerón
Estudiante de licenciatura
Título: gLV, can it find the essential?
Autores y afiliaciones:
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Abstract
Understanding the intricate relationship between interaction networks and ecosystem dynamics in ecological research is essential. We are dedicated to unraveling the complex interplay of these elements through simulating community dynamics. In this poster, we present our ongoing efforts to investigate how the properties of interaction networks shape and influence the ecosystems they inhabit.
While we are currently in the early stages of development, we are actively exploring ODE solvers to enhance the precision of our simulations. Additionally, we plan to implement neural networks in the future to identify network properties that predict the probability of species extinction and other community properties. These advancements will contribute to a deeper understanding of the intricate dynamics within ecosystems, furthering our ability to decipher the underlying complexities of essential organisms and their interactions.

Póster #11
Itzy Daniela Pérez Alvarado
Estudiante de licenciatura
Título: Exploring the Relationship between Phylogenetic Relatedness and Growth Characteristics in Bacteria
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Abstract
An important question in ecology and evolution is to what extent related organisms perform similar ecosystem functions, since differences among closely related species may point to specific adaptations. We wanted to test if related bacteria behave similarly to each other than more distant species. We are trying to determine if there is a correlation between growth characteristics, genomic features (genome size, number of genes), and phylogenetic relatedness among a group of bacteria isolated from the rhizosphere.
We obtained the genomes from the strains from the IMG database, and with them, we calculated the gANI of the strain. (genomic average nucleotide identity). Finally, we reconstructed a phylogenetic tree using the UPGMA algorithm. With the bar charts and the phylogenetic tree, it's possible to see that there is similarity between some of the organisms that have close phylogenetic relatedness, but there are some

others that show no similarity at all. Our next step is to calculate the phylogenetic signal, which together with our current results will inform strain selection for constructing synthetic bacterial communities.

Póster #23
Natalia Estrada Hernández
Estudiante de licenciatura
Título: Determining the role of the comER gene in biofilm formation in Bacillus mycoides
Autores y afiliaciones:
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1 Laboratorio Internacional de Investigación sobre el Genoma Humano .

Abstract
Biofilm formation is important for Bacillus species because of its role in their survival and ecological adaptation. Bacillus mycoides is a gram-positive bacterium that has been isolated from the soil and rhizosphere and is characterized by its filamentous biofilm morphology. The genetic regulation of biofilm formation by B. mycoides is poorly understood; however, the high conservation of the genes that have been identified in closely related Bacillus species can provide valuable insights. The comER gene has recently been identified to play an important role in biofilm formation in Bacillus cereus, an orthologous gene is found in Bacillus sp. 105MF. Therefore, the aim of this work is to identify the role of comER in biofilm formation in B. mycoides through the knockout of this gene.

Póster #8
Marcos Ernesto Ramírez Ramírez
Estudiante de licenciatura
Título: ¿Podemos nacer de nuevo? Evolución del ADN: la era de la simulación in sillico

Abstract
Actualmente existe una explosión de secuencias genómicas de diversos organismos, los cuales abren las puertas a nuevas preguntas sobre la evolución de las especies. Proponemos identificar asociaciones entre la variabilidad mutacional y la adaptación de los seres vivos a su entorno. Nuestro objetivo es desarrollar un método para analizar patrones mutacionales, en relación con variables ambientales, evolutivas y moleculares, para determinar su papel en la adaptación de múltiples linajes evolutivos. Proponemos realizar simulaciones en un cluster capaz de paralelizar los análisis de datos y la simulación programada en código Python, usando redes neuronales con algoritmos de retropropagación y genéticos en genomas de diferentes especies y linajes con información de las condiciones ambientales en las que vivieron, para identificar la relación de las mutaciones adaptativas principalmente y las no adaptativas en todo el genoma. Se espera que hay patrones que se pueden identificar en el puente entre el efecto del ambiente y la variación mutacional, que pueden ser modelados para realizar simulaciones. Este estudio nos ayudará a tomar mejores decisiones para la conservación de especies; además, nuestros resultados serán informativos para la reconstrucción de la historia de las especies; específicamente en la reconstrucción en conjunto de lo conocido actualmente de las condiciones ambientales pasadas, y su papel en la especiación.

Paleogenomics and Evolutionary Biology Lab

Póster #14
Pablo E. Uribe Herrera

Estudiante de licenciatura
Externo al LIIGH
Título: Reconstrucción De lGenoma Mitocondrial Antiguo De Equus sp. Encontrados En El Sitio Santa Lucia, Edo. Mex.
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Abstract
Conocer la historia evolutiva de los caballos prehistóricos a partir de su ADN ha sido de gran interés, debido al particular origen evolutivo de este género y su dispersión a través del mundo. La mayoría de los estudios se han centrado en abordar el proceso y dinámicas de su domesticación. Las muestras arqueológicas de caballo normalmente recuperadas tienen una antigüedad de no más de 5,000 años y proceden de Eurasia, aunque algunas datan del Pleistoceno tardío. Si bien este hecho ha permitido abordar la domesticación del caballo, se han dejado fuera múltiples preguntas científicas de gran interés. Una de ellas es el estudio de la evolución de los caballos en América que, aunque existen algunos trabajos, son pocos teniendo en cuenta que Norteamérica es la cuna de la familia de Equidae.Los hallazgos paleontológicos recientes en el sitio de Santa Lucía abren la posibilidad de realizar estudios paleogenómicos con caballos en México. Aquí, reportamos la obtención de ADN de siete muestras de caballos prehistóricos de Sta. Lucía datadas entre 12-30 mil años. Los datos preliminares sugieren la presencia de ADN endógeno y la reconstrucción exitosa de dos genomas mitocondriales. La comparación de estos genomas con otras muestras, antiguas y contemporáneas, permitirá contribuir al entendimiento de la historia evolutiva de esta icónica especie en América y el mundo.

Póster #25
Rigoberto Padilla Bustos
Estudiante de doctorado
Título: Characterization of the evolutionary history of the Columbian mammoth in Mexico
Autores y afiliaciones:
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Abstract

During the Pleistocene, one of the most abundant, and academically and socially interesting, extinct megafauna species was the mammoth. Two species inhabited America: the woolly mammoth (M. primigenius) and the Columbian mammoth (M. columbi), with the latter being the only endemic species to the continent. Recent studies suggest that the Columbian mammoth resulted from hybridization between an ancestral lineage of Siberian woolly mammoths and possibly the steppe mammoth. However, more genetic data on the Columbian mammoth is needed to better understand its peopling into America, its demographic history on the continent, social structure, evolutionary origin, and intricate hybridization history with the woolly mammoth, as well as, to address the possible causes of its extinction. The discovery of several Columbian mammoth remains at two sites northwest of the Cuenca de México, Santa Lucía and Tultepec, recovered during the construction of the Santa Lucía airport near Mexico City, provides a unique opportunity to study this species in depth from a paleogenomic perspective. More than 64 mitochondrial genomes have been recovered to date. However, these data represent the evolutionary history of a single genomic locus. In this poster I will discuss how autosomal data is essential to unequivocally characterize the evolutionary history of the Columbian mammoth, and to disentangle the possible causes of its demise.

Evolutionary Systems Biology Lab

Póster #38

Athena Tamayo Luisce

Estudiante de licenciatura

Título: Cuantificar control considerando la dinámica de respuesta de sistemas biológicos a una perturbación

Autores y afiliaciones:

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Abstract

El control por retroalimentación es una propiedad emergente que le permite a sistemas biológicos mantener una o varias variables de interés en un rango funcional incluso cuando los sistemas son perturbados. Cuantificar esta propiedad facilita evaluar, comparar y comprender los mecanismos subyacentes, así como su diseño para aplicaciones en biología sintética. Control Ratio, conocido como CoRa, es una aproximación general y sistemática para cuantificar el control por retroalimentación de sistemas biológicos. Esta metodología, desarrollada por Gómez-Schiavon y El-Samad (2022), compara las soluciones deterministas en estado estacionario del sistema de interés, descrito por ecuaciones diferenciales ordinarias, con las soluciones de un sistema idéntico pero sin retroalimentación, tanto antes como después de aplicarles una perturbación. Sin embargo, es importante señalar que CoRa no considera la dinámica de la respuesta del sistema a dicha perturbación. Esta dinámica puede conllevar costos adicionales para los sistemas, como grandes desviaciones iniciales del rango funcional, transitorias pero posiblemente tóxicas, o un tiempo prolongado para regresar al estado deseado. En este proyecto, nuestra meta es ampliar la aplicabilidad de CoRa al considerar dicha dinámica al evaluar el control por retroalimentación de sistemas biológicos.

Póster #1

Diego Morales Martinez

Título: Stochastic Bistability VS Transcriptional Bursting

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Abstract

Stochastic bistability appears when a system with only one steady state in its deterministic model displays a bimodal distribution –characteristic of bistable systems– in its stochastic representation. On the other hand, transcriptional bursting is a bimodal distribution resulting from the existence of 2 states of gene expression: inactive –with low transcription rate- or active -with high transcription rate. In both cases, the bimodal distribution is actually an emergent property of the noise, when only one steady state is predicted from the deterministic model. To evaluate how the dynamics of these 2 processes differ from each other, we developed 3 models of genetic circuits with: (1) positive feedback, which can show stochastic bistability, (2) a regulated gene, that can display transcriptional bursting, and (3) a hybrid system with both mechanisms. The conditions and properties where each of these systems display bimodal distributions will be characterized and compared.

Póster #33

Emiliano Sánchez Escalante
Estudiante de licenciatura

Título: Evaluating control and the emergence of signature behaviours in biological systems

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Abstract

Control" is a dynamic system's property to detect changes in its environment, and make intentional changes to its processes in order to maintain a specific variable as close to a desired value as possible. This property depends on a system's structure; and altering its interactions yields changes in its capacity to exert control. Using CoRa, we can quantify this capability, accurately showing how this capability changes within a parameter space. The main objective of this work is to determine whether the way control varies within a parameter space is unique to a specific system, or if it can exist within a different system's parameter space. Creating a large dataset by randomising the parameters of different biological systems, and utilising six different subsets of this dataset to determine the cluster number in the set, "signature behaviours" are identified as the recurring grouping of data points across repetitions wherein the trend for control capacity change is well defined. We conclude that these signature behaviours are not unique to one system, instead being recurrent across different systems. Going forward, the ability to classify a system's behaviour will allow us to predict the performance of any given system, to determine similarities between systems, and to infer the workings of more complex systems.

Póster #22

Rodrigo Aguilar Diaz
Estudiante de licenciatura
Título: Mathematical approach to quantify biological control in presence of biochemical noise
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Abstract
Homeostasis is the ability of biological systems to maintain a stable and relatively constant internal state even in the face of perturbations. Feedback control is the emergent property responsible for homeostasis, allowing the system to evaluate its own state and send a corrective signal accordingly. We use CoRa (Control Ratio) to quantify the ability of a biological control mechanism to maintain the system of interest at its steady state value by mathematically comparing the steady states of the system of interest and an analogous system without feedback. However, in its current form, CoRa only evaluates the system's deterministic response. In reality, the events necessary for these systems to function occur in a stochastic and discrete manner, which we call biochemical noise, causing cell-to-cell variation and unpredictable results. This is why we aim to expand the CoRa approach to measure the control of a system while taking into account its intrinsic stochastic nature. In doing so, we expect that this new approach will help us evaluate the contribution and limitations of control mechanisms in the presence of noise, which is necessary to understand the dynamic and regulatory properties of such mechanisms in a cellular context.

Genome Evolution Lab

Póster #18
Ricardo Alonso Echavarría Solana
Estudiante de doctorado
Título: Analysis of Hybrid Vigor Induced by Temperature Gradients Through Experimental Evolution
Autores y afiliaciones:
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Abstract
Agave fermentations are home to microorganisms whose metabolic activity provoke the rise of temperature above environmental levels. Saccharomyces cerevisiae, S. paradoxus and its hybrids have been isolated from agave fermentations in high proportions. Given the fact that high temperature is a stressor for both growth and reproduction, the project's objective is to evaluate if interspecific hybrids from both species could show improved fitness in these conditions, in a hybrid vigor scenario. To test this, we are going to direct a 5-step thermotolerance characterization in strains of both species and their hybrids isolated from agave fermentations. The best and worst growers of each parental species will then be hybridized in 6 combinations including intraspecific crossings. These parental strains and the 6 de novo hybrids will then be subjected independently to an evolution experiment in temperature gradients with consecutive transfer cycles spanning 5 months total. The growth rate under increasing

temperature will allow us to assess the fitness of hybrids with respect to a parental baseline. DNA samples for each strain before and after evolution will be obtained and sequenced for genomic analyses to inquire for ploidy differences, recurrent mutations, and loss of heterozygosity, allowing us to propose possible genomic mechanisms coming into play during the induction of thermotolerance by evolution.

Póster #28

Iván Eduardo Sedeño Jiménez
Estudiante de doctorado

Título: Introgressions in S. cerevisiae Isolates Associated to Agave Fermentations

Autores y afiliaciones:
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Abstract
In the world, there is a great variety of foods and beverages that undergo a fermentation process at some point, ranging from internationally popular drinks like beer to endemic foods like kimchi. Often, fermentation is carried out by yeasts of the Saccharomyces family, especially the species Saccharomyces cerevisiae. This species harbors a lot of genetic variation, allowing it to adapt to different environments where fermentation takes place. It is worth noting that members of this family have the characteristic of forming inter-species hybrids which have an evolutionary advantage to the niche where it formed, furthermore, it has been previously reported that the Mexican agave yeast population has a considerable number of introgressions when compared against yeast from around the world.
In this work, we present the analysis of 142 strains of Saccharomyces isolated from agave fermentations in different parts of the Mexican territory. Coverage analyses were conducted to identify the species to which each strain belonged to and the existence of gene flow among family members was explored. We successfully identified the coexistence of S. cerevisiae, S. paradoxus, and hybrids between these two species in the Saccharomyces population associated to agave fermentation. Finally, we describe the quantity and nature of DNA fragments from S. paradoxus in a population of S. cerevisiae in agave.

Póster #29

Maria Fernanda Requena Romo
Estudiante de licenciatura

Título: Catalase overexpression in Ustilago maydis and its Implications on the Virulence

Autores y afiliaciones:
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Abstract

In the study conducted in "Enhanced Oxidative Stress Resistance in Ustilago maydis and Its Implications on Virulence", the aim was to increase Ustilago maydis' resistance to H2O2 through experimental evolution and to identify the variants involved in this adaptation.

After analyzing variants in the adapted strain, it was discovered that one of them entailed the amplification of approximately 150kb of the left arm of chromosome IX. Within the genes implicated, UMAG_11067, responsible for encoding catalase, an enzyme known for catalyzing the decomposition of H2O2.

To evaluate catalase's contribution to resistance, an overexpression mutant was constructed by inserting the UMAG_11067 ORF fused with a constitutive promoter into the IP locus. Evaluating its resistance to oxidative stress revealed that catalase plays a role in adaptation. However, The increase in fitness due to catalase overexpression was smaller than that of the adapted strain. This suggests that in the adapted strain there are other contributing factors to H2O2 resistance.

Furthermore, the impact of catalase overexpression on virulence was studied by infecting maize seedlings. Results showed milder symptoms or symptom-free plants in the adapted strain and the catalase overexpression mutant, suggesting catalase may reduce virulence.

Further studies are needed to elucidate additional factors contributing to the adaptation and a more in-depth exploration of the catalase's role in virulence.

Póster #21

Maritrini Colón-González

Posdoctorante

Título: Using Yeast to Study Human Gene Variants Associated with Mitochondrial Disorders

Autores y afiliaciones:

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Abstract

Mitochondrial diseases (MD) are diverse disorders caused by mutations in mitochondrial or nuclear DNA. Over 1,500 nuclear genes impact mitochondrial structure and function, with about 27% associated with MD. Whole-genome and exome sequencing have uncovered new gene variants in MD, but not all are pathogenic. Saccharomyces cerevisiae, an ideal model, helps analyze these gene variants' impact on metabolism. This project aims to use S. cerevisiae systematically for analyzing human pathogenic gene variants.

As a proof of concept, we examined the human gene TUFM and its S. cerevisiae counterpart, TUF1. TUFM encodes Elongation Factor EF-Tu, crucial in mitochondrial protein synthesis. In 2019, a patient with MD exhibited a new single nucleotide variant in this gene.

We assessed the single nucleotide variant and complete knock-out impact on S. cerevisiae metabolism by comparing growth in fermentable and non-fermentable carbon sources. Findings reveal the knock-out mutant has modest growth delay under fermentative conditions, compared to the wild-type and single nucleotide substitution mutant. However, under non-fermentative conditions, the knock-out mutant's growth was impaired. Ongoing assessments involve evaluating mitochondrial function through membrane potential measurement, with additional analyses in progress.

Population and Evolutionary Genomics

Póster #36

Walter Nicolás Ortega
Estudiante de doctorado

Título: Genomic capture of HLA in individuals from the pre-hispanic and colonial period of Mexico

Autores y afiliaciones:
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Abstract
As a consequence of the Spanish conquest and colonization the population of Indigenous people was significantly reduced by epidemics during this period, estimating a decline of over 70% between 1520 and 1580.
One plausible factor contributing to the profound reduction in the Indigenous population may be linked to a weakened response of the adaptive immune system against pathogens introduced from distant regions. To investigate this, we aim to scrutinize the genetic diversity of the Human Leukocyte Antigen (HLA) system from 39 ancient individuals (34 from the colonial period and 5 from the pre-Hispanic period) to genomic capture using myBaits probes. Remarkably, we observed a substantial enhancement in the coverage and depth across 26 genes within the HLA region, achieving 100% coverage and depths approaching 25x in the best libraries.

Póster #7

Miriam Bravo López
Posdoctorante

Título: Reconstruction of one Salmonella enterica Paratyphi C genome from 19th-century Mexico City

Autores y afiliaciones:
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Abstract
The European colonization of Mexico resulted in devastating outbreaks that caused the death of millions of Indigenous individuals. To gain insights into the infectious agents introduced during this period, we generated paleogenomic data from seven individuals dated to the 19th century from Mexico City. Taxonomic classification of the obtained sequences enabled us to identify ancient DNA recovered from

one individual as Salmonella enterica Paratyphi C. This pathogen causes paratyphoid fever in humans. Remarkably, S. Paratyphi C has been proposed as one of the causative agents of the 1545-1550 cocoliztli epidemic, resulting in an estimated 15 million deaths. To better understand the evolutionary history of S. Paratyphi C in ancient Mexico, we reconstructed its genome at a 10x depth using an in-house capture-enrichment strategy. The reconstructed genome was phylogenetically analyzed alongside ancient and modern genomes of S. Paratyphi C. Our reconstructed genome falls within a branch closely related to the ancient S. Paratyphi C genomes from south Mexico. The divergence time between them was estimated to 500 years BP, which coincides with the Spanish conquest. Altogether, this study demonstrates that a replacement occurred within S. enterica Paratyphi C after the arrival of the Spanish conquistadors in New Spain.

Póster #4

María José Rodríguez Barrera & María Fernanda García Rodríguez
Estudiantes de licenciatura

Título: Genomic Analysis of ancient individuals from Quebec, Canada

Autores y afiliaciones:
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4 Licenciatura en Ciencias Genómicas, Escuela Nacional de Estudios Superiores Juriquilla.

Abstract
The archaeological site of Natashquan, in Canada, holds significant cultural and historical importance for the First Nation Natashquan community. However, since European contact, people from other regions have settled in the territory. This research focuses on the examination of ancient DNA (aDNA) obtained from archaeological samples from individuals excavated at the site, aiming to determine the potential origins of these ancient individuals and their genetic affinities.
Our study seeks to contribute valuable insights into the historical connection of the Natashquan community to the site, enhance understanding of Indigenous populations in North America, and support efforts to reclaim and preserve the cultural heritage of the region. This includes evaluating the possibility of restoring the land to its rightful Indigenous owners, the Natashquan community.

Póster #16

Anahí Tania Sánchez León

Título: Análisis paleogenómico de virus en el periodo prehispánico y colonial de México

Autores y afiliaciones:
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Abstract
La presencia de enfermedades infecciosas durante el periodo prehispánico y colonial en México se evidencia en el registro documental y arqueológico, lo que sugiere la existencia de diversos patógenos desde esa época. La colonización de México propició la transmisión de virus patógenos, lo que generó una crisis en la población indígena que se vio afectada por diversas epidemias. La paleogenómica ha

permitido la reconstrucción de genomas antiguos virales de DNA y RNA presentes en los restos arqueológicos. En este sentido, nuestro grupo de investigación, en un estudio previo, logró reconstruir el genoma de Hepatitis B y Parvovirus Humano (B19) en individuos del periodo colonial de México. Por lo tanto, este proyecto se enfocará en identificar y analizar genomas de virus humanos en un mayor número de individuos—siete individuos prehispánicos y 106 coloniales de México—a través de la asignación taxonómica con el programa MALT y con una base de datos actualizada que cuenta con 4,129 genomas virales. Se seleccionarán aquellos individuos con una mayor abundancia de secuencias virales antiguas para posteriormente, capturar y enriquecer su genoma. Este proyecto se suma a los primeros esfuerzos en identificar y caracterizar genomas virales antiguos, lo cual contribuirá al entendimiento del origen e historia evolutiva de los virus que circularon antes y después de la intervención española en México.

Póster #13

Barbara Beatriz Moguel Rodríguez
Posdoctorante
Population and Evolutionary Genomics, Paleogenomics and Evolutionary Biology, Externo al LIIGH

Título: Recovery of Sedimentary Ancient DNA from the Archaeological Site of Santa Lucia, Edo. México

Autores y afiliaciones:
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Abstract
The study conducted aimed to explore the microbial ecology of ancient environments, particularly focusing on sediment samples collected from Santa Lucia, Zumpango, Estado de Mexico. A total of 10 sediment samples were collected, and DNA extraction was performed to analyze microbial diversity. Subsequently, eight libraries were constructed, and sequencing was carried out, followed by taxonomic classification of the reads. Preliminary findings revealed the presence of two potentially pathogenic bacteria: Klebsiella pneumoniae and Mycobacterium canetti, detected in the oldest sediment layers retrieved. While M. canetti is closely related to Mycobacterium tuberculosis, the causative agent of tuberculosis (TB), it is exceptionally rare and thought to represent an ancestral form of TB-causing mycobacteria. On the other hand, K. pneumoniae, belonging to the Enterobacteriaceae family, can cause various infections in humans, such as pneumonia, bloodstream infections, and meningitis. Further validation analyses are necessary to confirm the authenticity of these findings and distinguish them from environmental relatives of the identified pathogens. This discovery marks a significant step in isolating pathogenic bacteria from ancient sediments, providing valuable insights into microbial ecology in ancient environments in Central Mexico.
Environmental paleomicrobiology offers promising avenues for studying microbial genome evolution processes and microbial ecosystem responses.

Mendelian Genomics and Precision Health Lab

Póster #5

Mariana Cuevas Bravo & Cassidy Guadalupe Luna Ruiz
Estancia académica

Título: Modelado funcional in vivo de enfermedades neurológicas humanas en Caenorhabditis elegans

Autores y afiliaciones:
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Abstract
Las enfermedades neurológicas son condiciones que afectan el sistema nervioso central, periférico y/o autonómico. Aproximadamente 15-20% de la población mundial vive con alguna condición neurológica que afecta su calidad y esperanza de vida. La mayoría de enfermedades neurológicas tienen un componente genético importante por lo que es esencial investigar sus causas moleculares, mecanismos y genes asociados. El desarrollo de plataformas experimentales utilizando modelos biológicos genéticamente manejables que permitan estudiar mecanismos de enfermedades es de gran importancia para entender los procesos biológicos involucrados en ellas.
El nemátodo Caenorhabditis elegans (C. elegans) representa un sistema ideal para estudiar enfermedades neurológicas humanas. Éste es un gusano de vida libre y fácil propagación en laboratorio; es transparente, lo cual permite la visualización de procesos celulares in vivo y se cuenta con herramientas para el estudio detallado del sistema nervioso. C. elegans tiene un genoma conocido con ~60% de genes ortólogos con el humano. Además se conoce su conectoma completo permitiendo reproducir fenotipos característicos de enfermedades neurológicas de interés. Por lo anterior, hemos desarrollado y establecido una plataforma de modelado in vivo utilizando C. elegans para estudiar los efectos y mecanismos de genes asociados a enfermedades neurológicas, incluyendo desórdenes del neurodesarrollo, neuromusculares, epilepsias y enfermedades neurodegenerativas.

Póster #24
Valeria Gómez Vela
Estudiante de licenciatura
Título: Genomic findings and SHANK 3 microduplication in a patient with autism spectrum disorder
Autores y afiliaciones:
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Abstract
Exome sequencing (ES) and chromosome microarray analysis (CMA) are genomic testing technologies to identify the molecular causes of heterogeneous neurodevelopmental disorders and identify pathogenic variants in patients with rare undiagnosed genetic disorders.
We report a 14 year old male patient referred for clinical features of autism spectrum disorder (ASD), language disorder and hyperkinetic disorder.
We performed trio exome sequencing in the affected proband and both unaffected parents and CMA in the proband in order to identify the molecular cause of the patient's phenotype. We identified a 13.363 kb microduplication in the 22q13.33 region encompassing the SHANK3 gene and a 11.643 kb

microdeletion in the Xq28 region affecting one exon of the GRABA3 gene. Additionally, ES identified a de novo predicted pathogenic novel splicing altering variant in the gene SMAD2.
SHANK3 encodes a scaffolding protein that plays important roles in the formation, maturation and maintenance of synapses. Phelan Mc-Dermid syndrome (PMS) is a rare genetic condition caused by loss of the SHANK3 gene through deletion of the 22q13 region or by pathogenic loss-of-function variants in the gene. Symptoms reported in patients with 22q13 duplication syndrome include mild developmental delay, behavioral problems and attention-deficit/ hyperactivity disorder (ADHD). The clinical presentation of our patient is consistent with the features reported in 22q13 duplication syndrome patients.

Póster #3
Marlon Aldair Arciniega Sánchez
Estudiante de licenciatura
Título: Genetic prevalence of early-onset and inherited dementias in the Mexican population.
Autores y afiliaciones:
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Abstract
Dementia is a worldwide public health concern affecting around 55 million people in the world, with over 60% living in low and middle-income countries. With the increasing proportion of elderly population in nearly every country, this number is expected to rise to 78 million by 2030. Early-Onset Alzheimer's disease (EOAD) and Frontotemporal Dementia (FTD) are known to be two of the main causes of dementia developing before 65 years old. FTD is the major cause of dementia in mid-life, whereas EOAD comprises 4%~5% of the total cases of Alzheimer's Disease, the major cause of dementia worldwide.
We aim to identify Pathogenic and Likely Pathogenic variants in five disease-associated genes categorized into two groups: GRN and MAPT for Frontotemporal Dementia (FTD), and PSEN1, PSEN2, and APP for Early-Onset Alzheimer's Disease (EOAD). Specifically, we are leveraging the Mexico City Prospective Study (MCPS) data to estimate the prevalence of these types of dementias in the Mexican population. The preliminary results of the study have identified six previously reported pathogenic and likely pathogenic variants in ClinVar associated with FTD and two known pathogenic and likely pathogenic variants associated with EOAD in individuals from MCPS.

Statistical Genomics of Complex Disease Lab

Póster #12
Cris Van Hout
Jefe de grupo
Título: Identifying Metabolically Healthy Obese Individuals Using R in the Mexico City Prospective Study.
Autores y afiliaciones:
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Abstract
This study investigated the paradoxical Metabolically Healthy Obese (MHO) phenotype using the Mexico City Prospective Study dataset capturing biometric and metabolic variables for a Mexican cohort. A comprehensive methodology was developed using R to reduce demographic and batch effects on metabolomic data like lipid and glucose levels, fluid balance, and inflammation indicators. Non-predictive factors for Body Mass Index (BMI) were omitted to focus on critical metabolic health biomarkers. A subset of high BMI individuals without typical obesity-associated dysfunctions was identified, indicating MHO presence. Unique MHO metabolic signatures were found by interpreting fitted residuals from regression models. Hierarchical modeling assessed metabolomic and demographic influences on metabolic health. Results place MHO individuals in the first quadrant of the metabolomic measures plot, with higher BMI than predicted by metabolomics. This highlights the need to further study these metabolic profiles to refine predictive models for metabolic disorders and nuance understanding of obesity.

Computational Population Genetics

Póster #34
Sebastián Iturbe
Estudiante de licenciatura
Título: Leveraging the Ancestral Recombination Graph to estimate the STR mutation model
Autores y afiliaciones:
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Abstract
Short tandem repeats (STR´s), also known as microsatellites are repetitive base pair sequences which comprise approximately 3% of the human genome. More than 10,000 STR variants have a contribution to gene expression and encompass 10-15% of the heritability of gene expression highlighting their role in the expression of various clinical conditions. One important caveat to understand the phenotypic contribution of STRs is that we need to fully understand the evolution and the mutation of STR´s. To do this, we must infer the mutation model that better explains patterns of genetic diversity. The evolution of STRs's depends on the evolutionary history across the genome showing the relationships between all the samples which are ultimately represented on the Ancestral Recombination Graph (ARG). The development of new methods to infer the Ancestral Recombination Graph (ARG), such as Relate and tsinfer, opens the door to the development of methods to infer the more likely mutation model taking into account the evolutionary history represented on the ARG. We propose the development of a new maximum-likelihood method to infer the most probable mutation model using the information encoded in the ARG. We show that our method gives accurate inferences of the STR mutation model after performing exhaustive simulations and point to possible applications of our method using data from the 1000 genomes project.

Póster #2
Jesús Abad Guzmán López
Estancia académica
Título: Impact of natural selection on synonymous sites on genetic diversity measures
Autores y afiliaciones:

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Abstract

Synonymous mutations are changes in the DNA that do not modify the amino acid composition of the proteins produced by a particular region in the DNA; these mutations often occur in the third base of a codon due to the genetic code redundancy. Synonymous mutations are assumed to have a neutral or nearly neutral effect since there are no alterations in the amino acid sequence. Evidence has shown that a high frequency of synonymous sites are under strong selection in species such as Drosophila melanogaster. Here we perform extensive simulations with the software SLiM to analyze the impact of natural selection on various measurements of genetic diversity such as the site frequency spectrum (SFS) through data analysis performed on informatic tools such as Python and R. We discuss the impact of our results on measurements of genetic diversity on various species.

El comité organizador les desea que disfruten de los días académicos 2024, gracias por su confianza y apoyo.