DÍAS ACADÉMICOS LIIGH 2026

5 y 6 de febrero del 2026
Centro Académico
Cultural, UNAM
Campus Juriquilla

CENTRO ACADÉMICO CULTURAL

LIIGH UNAM

Contents

Comité organizador

Responsables	Actividad académica
Daniela Robles Espinoza	Coordinadora / Investigadora Principal
Lucia G. Morales Reyes	Investigadora Principal
Federico Sánchez Quinto	Investigador Principal
Diego Ortega Del Vecchyo	Secretario Académico / Investigador Principal
Iliana Martínez Hernández	Delegada Administrativa
Eglee Lomelín de Anda	Asistente de la Coordinación
Carina Uribe Díaz	Técnica Académica
Jair Santiago García Sotelo	Técnico Académico
Maritrini Colón González	Investigadora postdoctorante
Evelia Lorena Coss Navarrete	Investigadora postdoctorante
Alessandro López Hernández	Estudiante de Posgrado
Iván Eduardo Sedeño Jiménez	Estudiante de Posgrado
Valeria Gómez Vela	Estudiante de Licenciatura
Emiliano Ferro Rodríguez	Estudiante de Licenciatura

Objetivo de los Días académicos del LIIGH-UNAM

La **tercera edición de los Días Académicos del LIIGH-UNAM**, que se llevará a cabo los días **5 y 6 de febrero de 2026**, tiene como propósito principal **difundir las investigaciones desarrolladas en el Laboratorio Internacional de Investigación sobre el Genoma Humano**.

Durante estas jornadas se presentarán **conferencias representativas de todos los grupos de investigación activos**, en las que se compartirán los avances más recientes en sus respectivas áreas. Entre las líneas de trabajo que actualmente se desarrollan en el LIIGH destacan: **genómica de poblaciones, genética del cáncer, paleogenómica, biología de sistemas, medicina de precisión, estadística, bioinformática, ecología y evolución**.

Además, el programa incluye **dos sesiones de pósters académicos**, elaborados por estudiantes de distintos niveles de formación y por posdoctorantes que realizan actividades de investigación en nuestra entidad, fortaleciendo así la participación y el intercambio académico en todos los niveles.

Figure 1: Imagen representativa. Te esperamos para la Foto grupal el viernes 6 de febrero, 2026 a las 12:30 h

Programa académico

Jueves 5 de febrero, 2026

Horario	Descripción	Moderador
08:45	Entrega de Póster	Carina Uribe Díaz
09:00	Ceremonia de inauguración y bienvenida	Dra. Daniela Robles
09:20	Los incontables cuentos de los cromosomas (Dr. Agustín Bernardo Ávila Casanueva)	Dra. Lucía Morales
10:20	**Mendelian Genomics and Precision Health Lab (Dra. Claudia Gonzaga)**	**Dra. Claudia Gonzaga**
10:20	Allelic spectrum and prevalence of G6PD Deficiency in the Mexican Population	Aldair Henández
10:30	Molecular and Clinical Variation Spectrum of Weiss-Kruszka Syndrome	Tania Sepulveda
10:40	Modeling Dosage-Sensitive Genes Within a 9p13.3 Microduplication Associated with a Rare Neurodevelopmental Genomic Disorder	José Luis Tellez
10:50	**Sesión de Pósters 1 & Coffee Break**	**Carina Uribe**
12:20	**Regulatory Genomics and Bionformatics Lab (Dra. Alejandra Medina)**	**Dra. Alejandra Medina**
12:20	Clinical, psychosocial, and demographic factors affect decisions in people with SLE	Domingo Martínez
12:35	Single cell approaches for large-scale multiomic analysis in the framework of JAGUAR Project.	Diego Ramirez
12:50	Lunch	Iliana Martínez
14:30	**Evolutionary Systems Biology Lab (Dra. Mariana Gómez)**	**Dra. Mariana Gómez**
14:30	Bistability and Transcriptional Bursting: A Comparative Study of Noise-Driven Switching Dynamics	Diego Morales
14:45	Mathematical modeling of negative feedback in maintenance of telomere homeostasis	Victoria Lelis
15:00	**Cancer Genetics and Bioinformatics Lab (Dra. Daniela Robles)**	**Dra. Daniela Robles**
15:00	Metabolic and Lipid Alterations Induced by High-Fat Diets in MASLD Development	Claudia Gutiérrez
15:15	Understanding structural variation in acral melanoma through whole genome sequencing	Kenya Contreras
15:30	**Paleogenomics and Evolutionary Biology Lab (Dr. Federico Sánchez)**	**Dr. Federico Sánchez**
15:30	Mitogenomas de camellos prehistóricos de la Cuenca de México incrementan nuestro entendimiento de la historia evolutiva de los Camellos de América	Eduardo Arrieta
15:45	Explorando el microbioma de Mammuthus columbi mediante paleometagenómica	Santiago Rosas
16:00	Fin del primer día	Dra. Daniela Robles

Viernes 6 de febrero, 2026

Horario	Descripción	Moderador
08:45	Entrega de Póster	Carina Uribe
09:00	**Population and Evolutionary Genomics Lab (Dra. María C. Ávila Arcos)**	**Laura Carrillo**
09:00	Ancient Bacterial Disease and Viral Exchange from Holocene Pathogen Genomes in Patagonia	Florencia Alvarez
09:10	Ancient HLA Variation and Predicted Immune Responses During Epidemics in Colonial Mexico	Walter Nicolas
09:20	Evolutionary Dynamics of Red Complex Periodontal Pathogens in Colonial Mexico Revealed by Ancient DNA	Itzy Pérez
09:30	**Statistical Genomics and Population Health Lab (Dr. Christopher Van Hout)**	**Dr. Christopher Van Hout**
09:30	Characterizing metabolic health and risk of cancer mortality using Principal Component Analysis in the Mexico City Prospective Study	Jair Contreras
09:45	Factores demográficos y de estilo de vida que influyen en el IMC en MCPS	Diego Morales
10:00	**Genome Evolution Lab (Dra. Lucía Morales)**	**Iván Sedeño**
10:00	Analysis of hybrid vigor in temperature gradients through experimental evolution	Ricardo Echavarria
10:15	Allele frequency dynamics in metagenomes from agave fermentation	Renata Sandoval
10:30	**Sesión de Pósters 2 & Coffee Break**	**Carina Uribe**
12:00	**Ecology and Evolution Lab (Dr. Sur Herrera Paredes)**	**Natalia Said Muñoz**
12:00	Diet-related gut microbiome differences in Systemic Lupus Erythematosus patients and healthy controls.	Yetel Ramírez
12:15	Identifying ecological interactions mediated by biosynthetic gene clusters in the ocean microbiome	Andrea Zermeño
12:30	Foto grupal (afuera del CAC)	Dr. Federico Sánchez
12:45	Lunch	Iliana Martínez
14:25	**Computational Population Genetics Lab (Dr. Diego Ortega)**	**Dr. Diego Ortega**
14:25	The impact of directional selection on patterns of genetic and phenotypic variation on a continuous space	Valeria Cabrera
14:35	Reconstrucción Paleogenómica del Desierto Chihuahuense: Explorando la Historia Evolutiva a través del sedaDNA	Laura Figueroa
14:45	Analysis of a Possible Relaxation of Natural Selection in Human Populations	Alejandra Marmolejo
14:55	**Sesión Informativa: VieRnes de Bioinformática**	**Dra. Evelia Coss**
15:05	Premiación y clausura	Dr. Federico Sánchez
15:35	Evento Social	Emiliano Ferro y Valeria Vela

Evento social

Con el objetivo de fortalecer la convivencia y crear espacios de esparcimiento, se han preparado dinámicas variadas que invitan a la participación de todos. Desde el deporte y la competencia amistosa, hasta los juegos de estrategia y creatividad, cada actividad busca generar momentos de diversión y colaboración.

Horario	Nombre de la actividad	Ubicación	Responsable
15:45	Partido amistoso de Fútbol	Cancha del campus, junto al gimnasio del campus	Federico Sánchez (cubículo 206) / David Figueroa (cubículo 104)
15:45	Mario Kart Wii	Área de encuentro académico	Emiliano Ferro (cubículo 209) / Valeria Vela (cubículo 203)
15:45	Juegos de mesa	Aula 3	Pavel Salazar (cubículo 205)
15:45	Búsqueda del Tesoro	Entrada del LIIGH	Maritrini Colón González (cubículo 104)

Especificaciones

Partido amistoso de Fútbol: Cada persona debe traer su equipo deportivo, que incluya tacos o zapatos adecuados para fútbol, una playera blanca (para un equipo) y una playera azul (para el otro equipo). No olvides traer una botella de agua y protector solar. Recuerda que lo más importante es venir con ganas de disfrutar y pasar un buen rato con amigxs. ¡Nos vemos en la cancha!

Mario Kart Wii: Para esta aventura rápida y furiosa, tu pandilla debe tener 4 integrantes. Competirán 4 pandillas por copa, donde cada copa consta de 4 pistas y cada pandilla elegirá a un personaje que será el nombre que corre con ustedes. Cada integrante deberá participar en una pista (el equipo decide el orden de quien va primero). El mismo juego dará un puntaje que se va acumulando en cada pista, el que obtenga más puntos pasa a la siguiente ronda. No se requiere saber jugar, solo mostrar tu espíritu de adrenalina callejera en el asfalto que corre como adicción en la sangre. Y tú, ¿Estas listx para levantar la copa pistón con tus amigxs?

Juegos de mesa: ¡Tarde de Juegos de Mesa! La idea de la actividad es compartir juegos de mesa, convivir entre nosotros y disfrutar juntxs un rato agradable. No necesitas saber jugar: vamos a traer varios juegos y nosotrxs te los enseñamos. Y si prefieres, también puedes traer tus propios juegos. Esta es una actividad libre así que puedes llegar cuando prefieras, aunque hay cupo limitado por el espacio. ¡Anímate a venir, todxs son bienvenidxs!

Búsqueda del Tesoro: La actividad se realizará en equipos de 6 a 8 personas. Caminarás por el campus buscando lugares y objetos para obtener las respuestas a los acertijos. Cada equipo deberá designar a una persona con un teléfono móvil con cámara, para poder acceder a un formulario de Google Forms y desde ahí tomar una fotografía como evidencia de la respuesta; además de usarlo para registrar el tiempo que tarde el equipo en realizar la actividad. La duración estimada es 90 minutos a paso lento, ¡así que no olvides traer zapatos cómodos, gorra, bloqueador solar y una botella de agua!

Resumen de las Presentaciones orales

Mendelian Genomics and Precision Health Lab

Responsable: Dra. Claudia Gonzaga Jauregui

Allelic spectrum and prevalence of G6PD Deficiency in the Mexican Population

Presenter: Aldair Hernández Morales
Authors: Aldair Hernández-Morales (1, 2), Claudia Gonzaga-Jauregui (1)
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1: International Laboratory for Human Genome Research, Laboratorio Internacional de Investigación sobre el Genoma Humano, Universidad Nacional Autónoma de México.
2: Undergraduate Program in Diagnostic Biochemistry, Facultad de Estudios Superiores Cuautitlán, Universidad Nacional Autónoma de México.

Abstract:
Glucose-6-phosphate dehydrogenase deficiency (G6PDd) is the most common enzymopathy worldwide, affecting about 5% of the population. Protein alterations can reduce the antioxidant capacity of red blood cells, predisposing carriers of pathogenic variants associated with G6PDd to episodes of acute hemolytic anemia, severe neonatal jaundice, or chronic non-spherocytic hemolytic anemia in rare more severe cases. In Mexico, G6PDd has been reported as the most frequent condition detected by different neonatal screening programs, with an estimated average prevalence greater than 4%, but with a wide geographical distribution in different regions of the country. Despite this prevalence, limited information exists on the genetic prevalence and disease-associated variants for G6PDd in the Mexican population. Through bioinformatic analyses of whole exome sequence data from 141,046 individuals part of the Mexico City Prospective Study (MCPS), we estimated that about 8 out of every 10,000 Mexican individuals in this cohort carry a variant associated with G6PD deficiency. We identified 18 pathogenic and likely pathogenic variants, of which c.844G>C;p.D282H (G6PD Seattle), c.1063A>G;p.I355V, c.1375C>G;p.R459G (G6PD Utah) and c.193A>G;p.T65A (G6PD Mexico DF) were the most common. Thirteen of these variants had not been reported in the Mexican population, although 11 of them had been previously reported in other populations. Additionally, we identified two novel variants that are predicted to result in loss of function: one frameshift deletion and one splicing variant. A better characterization of the allelic spectrum and prevalence of G6PDd in the Mexican population is fundamental to understand it and inform the development of public health strategies for early detection of the condition through newborn screening programs and the adequate management of individuals affected or at risk of developing complications triggered by medications or other oxidative agents.

Molecular and Clinical Variation Spectrum of Weiss-Kruszka Syndrome

Presenter: Tania Sepúlveda Morales
Authors: Tania Sepulveda-Morales (1), Joel Cota Castro (2,3), Luis Fernandez-Luna (1), Irene Valenzuela Palafoll (4), Raymond C. Caylor (5), Wesley Patterson (5), Anna Childers (5), Guillaume Jouret (6), Mylène Donge (7), Marwan Shinawi (8), Chaya N. Murali (9,10), Pilar Magoulas (9,10), Nicole P. Safina (11), Shelby Romoser (11), Jaclyn Zamzow (11), Karen J Villarroel Gomez (11), Emilia Athanasiou (12), Marina Gavatha (13), Jaime Vásquez (14), Berrin Monteleone (14), Barbara Masotto (15), M. Mar Rovira-Remisa (15), A. Cisneros (16), Amy Crunk (17), Juliette J. Kahle (17), Farah A. Ladha (9), Carlos A. Bacino (9,10), James R. Lupski (9,10), Carolina I. Galaz-Montoya (3,18), Claudia Gonzaga-Jauregui (1)
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6: National Center of Genetics (NCG), Laboratoire national de santé (LNS), Dudelange, Luxembourg.
7: Department of Pediatric Neurology, Centre Hospitalier de Luxembourg, Luxembourg City, Luxembourg.
8: Washington University School of Medicine, St. Louis, Missouri, USA.
9: Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, Texas, USA.
10: Texas Children's Hospital, Houston, Texas, USA.
11: University of Iowa Healthcare, University of Iowa Stead Family Children's Hospital, USA.
12: Clinical Genetics, Archbishop Makarios III Hospital, State Health Services Organization, Nicosia, Cyprus.
13: Pediatric Neurology Department, Archbishop Makarios III Hospital, State Health Services Organization, Nicosia, Cyprus.
14: NYU Langone Health, New York, New York, USA.
15: Department of Clinical Genetics, Hospital Universitari Germans Trias i Pujol, Badalona, Spain.
16: Hematology Department, Institut Català d'Oncologia (ICO Badalona) - Hospital Germans Trias i Pujol, Barcelona, Spain, Institut de Recerca Contra la Leucèmia Josep Carreras (IJC).
17: GeneDx, LLC, Gaithersburg, MD, 20877, USA.
18: Centro Médico Dr. Ignacio Chávez ISSSTESON Hermosillo, Sonora, Mexico.

Abstract:
Weiss-Kruszka syndrome (WSKA) is a rare genetic disorder caused by pathogenic variants in the ZNF462 gene or deletion of the 9q31.2 chromosome region encompassing ZNF462. In order to expand on the molecular and phenotypic spectrum of this rare genetic disorder, we report a case series of 12 individuals, aged 3.5 to 49 years, from five countries. Individuals were evaluated through chromosomal microarray analysis (CMA) for CNV analyses, exome-based gene panels and clinical or research based exome sequencing. We identified four deletions [del 9q31.2q31.3 (3.04Mb); del 9q31.2 (1.62Mb); del 9q22.33q32 (15.39 Mb); del 9q31.1q31.3 (7.8 Mb)], four frameshift variants [c.6174_6175delCT p.C2059Lfs*7; c.3474_3475del p.V1160Rfs*22; c.1815_1816dup p.(N606Ifs*8); c.6339del p.(R2114Gfs*43)], three nonsense [c.3700C>T p.(Arg1234Ter); c.5755C>T p.Gln1919Ter; c.5755C>T p.Gln1919Ter] and one missense variant [c.5098G>A p.Ala1700Thr]. Additionally, we reviewed the reported pathogenic allelic spectrum of ZNF462 reported in the literature and clinical databases. In total, 94 simple-nucleotide variants have been described, encompassing 37 nonsense, 50 frameshift, four splicing, and three missense variants; furthermore two translocations and 14 deletions involving the gene have been reported, including those identified in this study. The major phenotypes observed in WSKA include ptosis (81.8%), developmental delay (74.2%), nose features (63.6%), mouth features (62.1%), arched eyebrows (54.5%), intellectual disability (51.5%) and ear features (50%). This syndrome should be suspected in individuals presenting with mild global developmental delay and suggestive craniofacial features including ptosis, downslanting palpebral fissures, arched eyebrows, among others. Altogether, 66 patients have been documented in the literature, including the patients described in this study. Considering the reported cases in ClinVar, there could be at least 131 patients with a molecular diagnosis of Weiss-Kruszka syndrome, for an estimated prevalence of 1 in 60 million people. The phenotypic and molecular characterization of patients with rare diseases at different ages across diverse populations increases the understanding and awareness about these understudied genetic conditions.

Modeling Dosage-Sensitive Genes Within a 9p13.3 Microduplication Associated with a Rare Neurodevelopmental Genomic Disorder

Presenter: José Luis Tellez Arreola
Authors: José Luis Téllez Arreola1, Jazmín Fuentes Becerril2, Claudia Gonzaga-Jauregui1.
Affiliations:
1: International Laboratory for Human Genome Research, Laboratorio Internacional de Investigación sobre el Genoma Humano, Universidad Nacional Autónoma de México, UNAM Capus Juriquilla, , Querétaro, Mexico.
2: Bachelor's Degree in Diagnostic Biochemistry, Autonomous University of Mexico State. Mexico.

Abstract:
Structural genomic rearrangements, such as deletions, duplications, or inversions that occur sporadically and involve several kilobases to megabases of genomic DNA, affecting multiple genes simultaneously, often lead to genomic disorders associated with developmental delays and intellectual disability, sometimes accompanied by dysmorphic features and congenital anomalies. The severity of these disorders is closely linked to the size, number of genes involved, and, importantly, the expression of a few key dosage-sensitive genes within the affected genomic segment. To dissect and identify genes responsible for the disorder's phenotypes, it is important to study them functionally using in vivo systems. Here we report a case of a 6-year-old boy with a 304,941 bp duplication at 9p13.3, presenting with global developmental and motor delays, dysmorphic features, rotated ears, and strabismus. The duplication was not inherited from either of his unaffected parents, and it was shown to have occurred de novo in the patient. The duplication encompasses ten genes, none of which have been associated with human diseases to date. In order to identify potential dosage-sensitive genes driving the phenotype in the patient, we set out to functionally model four of the genes, namely ANKRD18B, SPINK4, BAG1, and CHMP5, that are well conserved in the nematode, Caenorhabditis elegans. We performed overexpression modeling of the C. elegans orthologs (T28D6.4, agr-1, bag-1, vps-60) to study their phenotype and potential links to nervous system function and development. We generated two independent lines overexpressing the T28D6.4 gene, with one line showing locomotion impairments compared to wild-type animals. However, we could not produce lines for bag-1 and vps-60 due to toxicity at any dosage. Our findings suggest that T28D6.4 gene expression may contribute to the observed human phenotype. Animal models are important resources to study the function of uncharacterized, well-conserved genes potentially associated with human diseases.

Regulatory Genomics and Bioinformatics Lab

Responsable: Dra. Alejandra Medina Rivera

Single cell approaches for large-scale multiomic analysis in the framework of JAGUAR Project.
Presenter: Diego Ramírez Espinosa
Authors: Diego Ramírez-Espinosa (1), Jorge Suazo Victoria (1), Karla Paulina Guzmán (1), Evelia Lorena Coss-Navarrete (1), Alejandra Schäfer (1), Felipe Gajardo (2), Julieth Lopez-Castiblanco (3), Marcela Sjoberg (2), Yesid Cuesta-Astroz (3), Pablo Romagnoli (4), Danilo Ceshin (4), Tarran Rupall (5), Anna Lorenc (5), Carla Jones (5), Gosia Trynka (5), Alejandra Medina-Rivera (1).
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3: Instituto Colombiano de Medicina Tropical, Colombia.
4: Centro de investigaciones en Medicina Traslacional "Severo R. Amuchástegui" (CIMETSA), Instituto Universitario de Ciencias Biomédicas de Córdoba (IUCBC), Argentina.
5: Wellcome Sanger Institute, UK.

Abstract:
Single-cell assays are among the most powerful approaches for studying the biology of specific cellular populations at high resolution. Within the JAGUAR Project (Joining All: Genes, immUnity and diveRsity), we aim to generate the first deep atlas of immune cell composition in Latin American populations by integrating multiple single-cell technologies, including scATAC-seq, scRNA-seq, and CITE-seq. This strategy enables a comprehensive characterization of transcriptional states and regulatory programs shaping immune system diversity in these populations. Although numerous computational methods have been developed to analyze single-cell data, most are designed for individual modalities and operate within isolated, often incompatible analytical frameworks, limiting the ability to draw joint biological conclusions across technologies. While multi-modal assays such as Multiome or DOGMA-seq allow the simultaneous profiling of multiple molecular layers within the same cells, their high cost and frequent data quality limitations restrict their applicability, particularly for deep analyses of rare cell populations at large scale. Here, we present a pipeline in development that enables quality control, normalization, large-scale batch correction, and integration of multiple single-cell technologies within a unified analytical framework. Our approach combines complementary bioinformatic tools, including Signac, Seurat, PoissonVI, SCENIC+, and additional methods for multimodal modelling and demultiplexing, alongside multiple reference datasets for robust cell type annotation. We place particular emphasis on chromatin accessibility analysis, a modality that poses unique computational and statistical challenges at scale, and showcase results in cell type annotation, batch integration, and transcription factor analysis. Finally, we outline future analytical directions enabled by this framework, including the integration of expression quantitative trait loci (eQTLs) informed by the regulatory and expression variability captured in our analyses. This extension would add a functional genetic layer that can further link regulatory variation to immune phenotypes, enhancing the interpretability and translational potential of the atlas.

CLINICAL, PSYCHOSOCIAL, AND DEMOGRAPHIC FACTORS AFFECT DECISIONS IN PEOPLE WITH SLE

Presenter: Luis Domingo Martínez Vázquez
Authors: Hernández-Ledesma Ana Laura (2) †,Pedraza-Meza Luis Miguel(1,2) † ,Bravo-García María Fernanda(2) , Ruiz-Contreras Alejandra E (3), Medina-Rivera Alejandra(2)*, Martínez Domingo(2)*
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† These authors contributed equally to this work and share first authorship.

Abstract:
The Mexican Lupus Registry (LupusRGMX) was established to address the lack of population-specific data on systemic lupus erythematosus (SLE) in Mexico. By integrating clinical, demographic, and psychosocial information reported by patients, LupusRGMX provides a unique platform to characterize disease heterogeneity and to study how SLE affects multiple dimensions of patients' lives. The present study is entirely based on data from LupusRGMX and examines decision-making processes in people with SLE. The main objective was to evaluate how clinical, psychosocial, and demographic factors influence social and temporal decision-making. In addition, agreement between patient-reported and rheumatologist-assessed disease activity was assessed. Statistical analyses included McNemar and chi-squared tests, Bland–Altman plots, and regression models to evaluate systematic bias, agreement, and behavioral responses. Patient-reported remission states based on the SLEDAI score showed strong concordance with

rheumatologist assessments. Differences between patient and physician scores were clinically insignificant (<4 points), supporting the use of a binary remission classification. Moreover, discrepancies between scores decreased proportionally with decreasing disease activity, indicating greater agreement in patients with less active disease. Remission status and glucocorticoid use played a central role in shaping decision-making. Individuals with SLE in remission tended to impose higher punishment on social norm violators and preferred immediate rewards over larger, delayed rewards. Glucocorticoid use was also associated with a preference for immediate rewards, and higher doses were linked to increased punitive behavior. Psychosocial and demographic factors further modulated behavior: increased age and hostility were associated with greater punishment, while higher obsessive–compulsive traits were linked to impulsivity and preference for sooner rewards. Among all variables examined, remission status showed the largest effect on both social and temporal decision-making. These findings were replicated in an independent validation sample, underscoring the robustness of results derived from LupusRGMX.

Evolutionary Systems Biology Lab

Responsable: Dra. Mariana Gómez Schiavón

Bistability and Transcriptional Bursting: A Comparative Study of Noise-Driven Switching Dynamics

Presenter: Diego Morales Martínez
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2: Universidad Autónoma de Querétaro.
3: Millennium Science Initiative Program, Millennium Institute for Integrative Biology (iBio), Chilean National Agency for Research and Development, Santiago 8331150, Chile.

Abstract:
At molecular scales, biological systems are stochastic and this randomness is known as biochemical noise. In the presence of biochemical noise, positive feedback with bistability and transcriptional bursting models exhibit bimodal distributions and switch dynamics; in both cases, these are actually emergent properties of the noise. By analyzing their dynamical properties and noise-driven transitions, we aim to discover how these mechanisms contribute to cellular heterogeneity. In particular, we seek to determine whether there are biologically relevant differences between the two mechanisms—for example, whether a gene regulatory network with positive feedback confers higher fitness than an otherwise identical network driven by transcriptional bursting. Altogether, this comparative framework is intended to assess whether distinct noise-driven regulatory mechanisms can produce similar phenotypic outcomes or instead differentially impact robustness, adaptability, and fitness, despite generating comparable levels of cellular heterogeneity. To evaluate how the dynamics of these two mechanisms differ, we developed three models of genetic circuits: (1) a positive feedback model, which can exhibit bistability, (2) a regulated gene that can display transcriptional bursting, and (3) a hybrid model incorporating both mechanisms. The conditions and properties under which each of these mechanisms displays bimodal distributions will be characterized and compared. As preliminary results, we found that in the deterministic representation the transcriptional bursting model is monostable under all conditions. While the positive feedback and hybrid model depends on the parameter values for determining its global dynamic. In the latter two cases, we observed different behaviors.

Mathematical modeling of negative feedback in maintenance of telomere homeostasis
Presenter: Victoria Rodríguez Lelis
Authors: Victoria Rodríguez Lelis [1, 2], Mariana Gómez Schiavón [1, 3]
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2: Escuela Nacional de Estudios Superiores Juriquilla, UNAM.
3: Millennium Science Initiative Program, Millennium Institute for Integrative Biology (iBio), Chilean National Agency for Research and Development, Santiago, Chile.

Abstract:
Telomeres are repetitive DNA sequences located at the ends of chromosomes, essential for protecting genomic integrity. Maintaining an optimal telomere length is crucial for this function. Critically short telomeres trigger DNA damage responses and genomic instability. Conversely, excessively long telomeres—often sustained by persistent telomerase activity—are associated with cellular immortalization and cancer. The regulation of telomere length involves a dynamic interplay between the telomerase enzyme and the shelterin protein complex. Here we propose that a negative feedback loop, mediated by shelterin complexes and telomerase, constitutes the core mechanism for sustaining telomere length homeostasis. To explore this hypothesis, we developed a simple mathematical model based on a regulatory mechanism where the number of shelterin complexes bound to a telomere is length-dependent. In this model, long telomeres accumulate more shelterin complexes, which strongly inhibits telomerase binding and prevents telomere elongation. Conversely, short telomeres, with fewer bound shelterin complexes, present minimal inhibition, permitting telomerase access and subsequent elongation. Through dynamical analysis of the model, we aim to characterize the system's homeostatic properties and evaluate how genetic perturbations (e.g., mutations in shelterin complex or telomerase) can disrupt stability. This work provides a theoretical framework to explore how negative feedback governs homeostatic capacity in telomere length regulation, thereby elucidating its robustness and failure modes.

Cancer Genetics and Bioinformatics Lab

Responsable: Dra. Daniela Robles

Metabolic and Lipid Alterations Induced by High-Fat Diets in MASLD Development
Presenter: Claudia Gutiérrez García
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Abstract:
Metabolic dysfunction-associated steatotic liver disease (MASLD) is a chronic liver disease that

can evolve into irreversible pathologies such as cirrhosis and hepatocellular carcinoma. More than a third of adults worldwide are affected by it, and its prevalence is projected to keep increasing over the following decades. One of the main contributing factors to develop MASLD is high-fat diets, which makes it reversible. This work aims to investigate how high-fat diets modify the zonation of lipid metabolism and their effect on the circadian cycle. Zonation is a structural and functional organization of cells into distinct areas within the same tissue. The circadian cycle refers to the regulatory system that coordinates physiological, metabolic, and behavioral processes in a rhythmic manner. We designed an experimental mouse model consisting of two high-fat diets (one containing 10% fat and the other 40%) administered through different stages of MASLD development. Zonation is studied through the staining of specific enzymes in collected liver tissue, and the circadian cycle is analyzed through metabolic cages. We have conducted the first experiments from our experimental design, observing the following results. For the 10% fat diet group, we preliminarily observe a shift in the zonation of the enzyme glutamine synthetase on day 4, as well as circadian cycle alterations. For the 40% fat diet group, hepatic steatosis was observed on week 16, which is consistent with an increase in total cholesterol and a decrease in triglyceride levels in blood, including higher disruption of circadian cycle. Our preliminary findings indicate that high-fat diet consumption induces hepatic steatosis contributing to MASLD development by altering hepatic zonation, lipid metabolism at both tissue and systemic levels, and the circadian cycle.

Understanding structural variation in acral melanoma through whole genome sequencing

Presenter: Kenya Lizbeth Contreras Ramírez

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Abstract:

Background. Melanoma is the most dangerous type of skin cancer. Acral melanoma (AM) is a subtype of melanoma that arises on the palms of the hands, soles of the feet and under the nails. Unlike cutaneous melanomas, AMs show high rates of complex genomic rearrangements involving hundreds to thousands of structural variants (SVs). AM is the most common subtype of melanoma in Mexico and other countries in Latin America, Africa and Asia, and represents a small fraction of melanoma cases in people of European ancestry. AM patients respond poorly to current treatment strategies, and, due to the scarce research on this type of cancer, no specific therapies are available for them. Its causes are still unknown, and the relative contribution of genetic ancestry, environmental factors, and tumor biology to its aggressive behavior remain unknown. Consequently, understanding the landscape of structural variation in acral melanoma through whole-genome sequencing (WGS) provides insight into the biological mechanisms underlying its genomic complexity and aggressive evolutionary behavior. Methods. We have generated WGS data for 21 tumor–normal sample pairs from Mexican AM patients, and we included 87 previously published samples from Australian patients (Newell et al., 2020). We

have leveraged a unique suite of methods developed and used at the Cortés-Ciriano lab for the study of cancer genome evolution. These methods include functionalities for the alignment and processing of sequencing reads, detection of diverse types of somatic mutations (SNVs, SVs, and copy number aberrations), and classification of complex genomic rearrangements. Preliminary results. So far, we have called SV, copy number alterations, and complex rearrangements, and we identified specific patterns in 32% of patients for which we propose a hypothesis of tumor evolution mechanisms, including chromothripsis and breakage-fusion-bridge cycles. We are also exploring additional structural events that may reveal alternative evolutionary mechanisms. Conclusion. We have confirmed characteristics previously reported in other studies, including those from our own group, and our analyses are enabling the stratification of patients according to their genomic profiles and possible mechanisms of tumor evolution.

Paleogenomics and Evolutionary Biology Lab

Responsable: Dr. Federico Sánchez Quinto

Mitogenomas de camellos prehistóricos de la Cuenca de México incrementan nuestro entendimiento de la historia evolutiva de los Camellos de América

Presenter: Eduardo Alejandro Arrieta Donato
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Abstract:
La primera aparición en el registro fósil del género Camelops (camellos prehistóricos) se remonta desde hace 4 millones de años en Norte América hasta su extinción hace 11,000 años, al final del Pleistoceno. Un reciente estudio sobre mitogenomas de Camelops hesternus del Pleistoceno tardío de Yukón (Canadá), reportó que esta especie es más cercana genéticamente a los camellos contemporáneos de África y Asia, que a especies de camélidos de Sudamérica, como las llamas o vicuñas. Sin embargo, los Camelops habitaron Norte y Centro América, por lo que no es claro si los datos genéticos de los camellos de Yukón representan la variación genética mitocondrial completa de los camellos de ésta especie a lo largo del continente americano. El reciente descubrimiento de restos de camellos prehistóricos asociados al Pleistoceno tardío en el área conurbada de la Ciudad de México, proporciona una oportunidad única para el estudio de la historia evolutiva del género Camelops en América. En éste estudio se extrajo, secuenció y analizó DNA antiguo de siete muestras de camellos. Se maximizó la información genética de tres muestras a través de experimentos de enriquecimiento-captura, recuperando tres mitogenomas de camellos a más de 9x de cobertura. Los distintos análisis filogenéticos efectuados con estas tres muestras mexicanas en conjunto con seis muestras del mismo género de Canadá y E.E.U.U, y camellos contemporáneos, revelaron una profunda estructura y dos principales clados de camellos antiguos en Norte América. Nuestros resultados podrían ser explicados por

eventos migratorios de gran escala geográfica, los cuales forjaron esta peculiar estructura en la variación mitocondrial de camellos en el continente. Este estudio es fundamental para el entendimiento de la historia evolutiva de los camellos en América, particularmente, debido al incorporamiento de muestras antiguas procedentes de latitudes subtropicales.

Explorando el microbioma de Mammuthus columbi mediante paleometagenómica

Presenter: Santiago Rosas Plaza
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Abstract:
Los estudios sobre la interacción hospedero–microbioma han demostrado que los macroorganismos albergan una alta diversidad de microorganismos con los que establecen interacciones complejas, incluyendo simbiosis e incluso filosimbiosis. Los avances en paleogenómica han permitido extender el estudio de estas interacciones al pasado mediante el análisis del microbioma y de microorganismos específicos que han coexistido con los hospederos durante miles de años. Hasta el momento, los estudios paleogenómicos del microbioma se han enfocado principalmente en el ser humano, utilizando coprolitos para el estudio del microbioma intestinal y sarro dental para el microbioma oral. A pesar de que existen restos paleoarqueológicos con potencial para el estudio del microbioma de otros macroorganismos, estas aproximaciones han sido poco exploradas fuera del contexto humano. El objetivo de este trabajo es ampliar esta línea de investigación mediante el análisis del microbioma oral de Mammuthus columbi. Se analizaron muestras de dentina de 77 mamuts del Pleistoceno, recuperadas en la Base Aérea Militar de Santa Lucía y en Tultepec, México. Todas las muestras fueron sometidas a secuenciación shotgun y a análisis metagenómico con Kraken2. A partir de estos resultados, se seleccionaron grupos microbianos relevantes para realizar mapeos genómicos contra genomas de referencia modernos para evaluar patrones de daño característicos de ADN antiguo. Posteriormente, se realizaron análisis filogenéticos utilizando genomas de cepas modernas aisladas de mamíferos y cepas recuperadas de mamuts para investigar las relaciones entre cepas antiguas y modernas. La detección de potenciales miembros del microbioma oral de M. columbi resalta la importancia de la paleometagenómica como una herramienta clave para el estudio de las interacciones hospedero–microbioma en hospederos extintos. En este contexto, la ampliación de estos estudios a otros miembros de la megafauna extinta y su comparación con parientes actuales podría contribuir a profundizar en procesos de coevolución hospedero–microbioma y posibles patrones de filosimbiosis.

Population and Evolutionary Genomics Lab

Responsable: Dra. María C. Ávila Arcos

Ancient Bacterial Disease and Viral Exchange from Holocene Pathogen Genomes in Patagonia

Presenter: Florencia Alvarez Gallego
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Abstract:
Patagonia was the last mainland territory to be peopled by humans around 14,000 years ago. The recovery of ancient pathogen genomes from this region as old as the 6000 yBP offers a valuable perspective to explore the local impact of infectious diseases in the past and their evolutionary processes. This study recovered ancient DNA from 32 individuals from Central Patagonia dating around 6000-100 yBP. We generated paleogenomic data, performed metagenomic characterization, and recovered one Hepatitis B virus genome (6.8 X, 1,500 yBP) and four genomes of the second leprosy-causing pathogen Mycobacterium lepromatosis (16.2-1.1X, 4,000–100 BP). We examined ancestral recombination events in extinct ancient HBV lineages from South America that belong to an Ancient American clade. This analysis identified ancient American genomes that possibly acted as parents and may have contributed to the diversification of modern H and F genotypes. Our recovered ancient HBV genome allows a new calibration to point for estimating the recombination rate and lineage split times. In parallel, we analysed ancient M. lepromatosis genomes, representing the oldest and southernmost lineages recovered to date. Their phylogenetic placement indicates an affinity with human-associated strains, suggesting a millennia-old pathogenic relationship between humans and M. lepromatosis and adding additional calibration points to date the divergence from M. leprae, leprosy's first causing pathogen. Together, these results provide an evolutionary perspective on the dynamics in ancient pathogens native to the Americas.

Ancient HLA Variation and Predicted Immune Responses During Epidemics in Colonial Mexico

Presenter: Walter Nicolas Ortega
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10: Department of Ecology, Evolution, and Organismal Biology, Brown University, U.S.A.

Abstract:
During the colonial period in New Spain, interactions among Indigenous, European, and African populations led to profound demographic and genetic changes. Between 1520 and 1580, successive epidemics caused a dramatic decline in the Indigenous population, with mortality exceeding 80%. Differences in adaptive immunity, particularly within the human leukocyte antigen (HLA) system, may have contributed to variation in survival during these outbreaks. To investigate HLA diversity and its potential impact on immune responses during colonial epidemics, we analyzed ancient DNA (aDNA) from 59 individuals, including 54 from the colonial period and 5 from pre-Hispanic contexts. HLA regions were enriched using myBaits in-solution capture followed by next-generation sequencing (NGS). HLA genotyping was performed using the OptiType and TARGT pipelines. In addition, NetMHCpan and NetMHCIIpan were used to predict the binding affinity of HLA variants to pathogen-derived peptides, providing insights into immune responses that may have influenced survival. Our results yielded high-quality data, achieving complete coverage of 26 HLA genes and an average sequencing depth of approximately 25x in high-quality libraries. We identified several HLA alleles, including HLA-A02:01, HLA-B35, HLA-C04:01, HLA-DPA101:03, HLA-DPB104:02, HLA-DQA103:01, HLA-DQB1*03:02, and HLA-DRB1*08:02. The frequencies of these alleles are similar to those observed in present-day Mexican populations. Furthermore, NetMHCpan and NetMHCIIpan analyses revealed HLA variants with strong predicted binding affinities to pathogen-derived peptides. To further evaluate the structural plausibility of these interactions, we performed molecular docking analyses of selected HLA–peptide complexes, which supported stable binding conformations and reinforced the potential immunological relevance of the predicted interactions during historical epidemics. Overall, this study provides valuable insights into the genetic factors that may have shaped survival outcomes during one of the most severe demographic crises in the history of the Americas.

Evolutionary Dynamics of Red Complex Periodontal Pathogens in Colonial Mexico Revealed by Ancient DNA

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Abstract:
The European colonization of Mexico triggered profound epidemiological transitions, including the introduction of novel microbial lineages. While ancient DNA (aDNA) studies have documented the colonial introduction of specific oral pathogens, such as certain lineages of Tannerella forsythia, the broader genomic dynamics of synergistic periodontal communities remain incompletely characterized. In periodontal disease, T. forsythia acts alongside Porphyromonas gingivalis and Treponema denticola, collectively known as the “red complex”, which drives pathogenesis. Therefore, characterizing these species in a colonial context is essential to understanding how colonial dynamics reshaped the oral microbiome. To explore this dynamic, we analyzed aDNA from teeth (n=96) and dental calculus (n=7) samples belonging to 96 individuals from two colonial-era sites in Mexico City: the Hospital Real de San José de los Naturales (HSJN) and the Templo de la Inmaculada Concepción (“La Conchita”). We performed DNA extraction, high-throughput sequencing, and bioinformatic analysis using Kraken2 to taxonomically classify non-human DNA and screen for ancient pathogens. Notably, we recovered members of the periodontal “red complex”, including Porphyromonas gingivalis, a key pathogen of periodontitis, and Treponema denticola. Preliminary phylogenetic analyses of colonial T. denticola strains reveal two major clusters: one comprising pre-contact strains and another associated with post-contact contexts. This separation suggests the introduction of novel T. denticola lineages during the colonial period, mirroring patterns observed in T. forsythia. In contrast, P. gingivalis exhibits the persistence of multiple lineages across both temporal contexts, indicating a more continuous pattern of diversity. Beyond evolutionary insights, this study shows how the arrival of new populations reshaped oral microbiomes in colonial Mexico. By integrating paleogenomics with historical context, we uncover how social transformations are recorded in microbial DNA.

Statistical Genomics and Population Health Lab

Responsable: Dr. Cris Van Hout

Characterizing metabolic health and risk of cancer mortality using Principal Component Analysis in the Mexico City Prospective Study
Presenter: Jair Emiliano Contreras Rivera
Authors: Cristopher Van-Hout(1), Jair Rivera (1), Lorena Gutierrez (2)
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Abstract:
Epidemiological studies linking cancer risk to obesity have traditionally relied on Body Mass Index (BMI) to characterize healthy and unhealthy metabolic profiles. However, BMI has important limitations, it does not fully capture metabolic heterogeneity and can overlook differences in risk among individuals with similar BMI values. This study aimed to characterize metabolic health among participants in the Mexico City Prospective Study (MCPS) using Principal Component Analysis (PCA) to integrate information from Nuclear Magnetic Resonance (NMR) spectroscopy based metabolomics measures and mortality data. After quality control, PCA was applied to 249 biomarkers and the first 10 principal components accounted for 90% of the total variance. These components were used to model BMI as a function of metabolic biomarkers, adjusting for relevant covariates. The fitted value, or predicted BMI, captures the extent to which an individual’s metabolic profile corresponds to higher or lower BMI and is used here as a measure of Predicted Metabolic Health (PMH). Associations between PMH, BMI, age, sex, and risk of BMI related cancers among 2,224 cases and 147,333 controls were then evaluated in a multivariable logistic model. In addition to established risk factors, PMH demonstrated a significant

independent association with risk of cancer mortality, with a greater effect size than BMI. These findings suggest that BMI alone is insufficient to characterize obesity related disease risk and highlight the value of multivariate approaches, such as PCA, in providing a more parsimonious and informative representation of metabolic profiles.

Factores demográficos y de estilo de vida que influyen en el IMC en MCPS
Presenter: Diego Ivan Morales Hernandez
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Abstract:
La Cohorte Mexicana de Estudios Poblacionales (Mexico City Prospective Study, MCPS) ofrece un amplio recurso para examinar los determinantes demográficos y de estilo de vida del Indice de Masa Corporal (IMC) y los resultados de salud relacionados. Utilizando datos de más de 140,000 participantes, evaluamos las asociaciones entre los factores de estilo de vida y el IMC en diferentes estratos de edad y sexo. Los patrones observados en MCPS son en general consistentes con los hallazgos de otras cohortes grandes, incluyendo el Biobanco del Reino Unido, pero sugieren efectos específicos de la edad y el sexo que no se han caracterizado adecuadamente en otros estudios. Los análisis indican que las distribuciones del IMC varían sistemáticamente según el grupo demográfico, con diferencias entre deciles de edad y entre hombres y mujeres. Factores de estilo de vida como el tabaquismo y la actividad física contribuyen a estos patrones; la actividad física se asocia con distribuciones de IMC más saludables, mientras que el tabaquismo muestra asociaciones más débiles. Es importante destacar que la agrupación de comportamientos como el consumo de alcohol, el tabaquismo y la actividad física resalta la necesidad de considerar simultáneamente múltiples dominios del estilo de vida. El trabajo futuro en MCPS ampliará los análisis para incluir la nutrición general, el sueño y factores socioeconómicos, e incorporará biomarcadores de adiposidad y función metabólica para fortalecer la inferencia. Los análisis longitudinales serán cruciales para desentrañar las vías causales y evaluar cómo interactúan los hábitos de vida a lo largo de la vida. MCPS ofrece una plataforma para avanzar en la comprensión del riesgo de obesidad y enfermedades crónicas en México y contribuir a la evidencia global.

Genome Evolution Lab

Responsable: Dra. Lucía Morales

Analysis of hybrid vigor in temperature gradients through experimental evolution
Presenter: Ricardo Alonso Echavarría Solana
Authors: Ricardo Echavarría Solana (1), Lucía Morales (1).
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Abstract:
Hybridization between closely related species is often associated with fitness trade- offs, such as infertility and genomic incompatibilities. However, it can also confer advantages that contribute to adaptive variation—a phenomenon known as hybrid vigor. In Mexico, traditional agave fermentations used for distillate production occur in open tanks, allowing microbial exchange between fermenting must and the environment. Here, Saccharomyces cerevisiae coexists with its sister species S. paradoxus and their hybrids, which occur at higher frequencies in semi-arid regions.

Notably, these species differ in thermotolerance—a critical trait for these fermentations. We hypothesize that the high prevalence of hybrids in these environments may result from enhanced thermotolerance relative to parental strains. To test this, we will conduct experimental evolution in temperature gradients using parental and de novo hybrid strains with differing thermotolerance profiles, characterizing both fitness dynamics and genomic changes linked to adaptation across 200 generations. So far, we have evaluated 28 sequenced agave yeast strains (including both species and hybrids) for growth at elevated temperatures (up to 42°C) in rich media. The best- and worst-performing strains were selected to generate new hybrids with diverse parental genotype-phenotype combinations, which will undergo experimental evolution.

Allele frequency dynamics in metagenomes from agave fermentation
Presenter: Renata Sandoval Chiw
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Abstract:
Agave fermentations used for the production of traditional distilled beverages in Mexico host complex microbial communities, in which Saccharomyces cerevisiae is commonly reported as a key producing microorganism. We analyzed a complete fermentation process, from initiation to completion, at Hacienda de Vergel Guadalupe, where we observed a contrasting pattern in which the bacterium Zymomonas mobilis was the most prevalent species. Using metagenome-assembled genomes (MAGs), we examined allele frequency changes across the fermentation timeline to characterize its evolutionary dynamics. Our preliminary results reveal a progressive accumulation of single-nucleotide polymorphisms (SNPs) throughout the process, suggesting ongoing genetic diversification. Future analyses reconstructing allele frequency trajectories will allow us to identify SNPs that may become fixed in the population and subsequently annotate their genomic and functional significance.

Ecology and Evolution Lab

Responsable: Dr. Sur Herrera Paredes

Diet-related gut microbiome differences in Systemic Lupus Erythematosus patients and healthy controls.
Presenter: Yetel Ramírez Mosqueda
Authors: Yetel Ramírez-Mosqueda (1)(2), María-Fernanda Bravo-García (1), Alejandra Medina-Rivera (1), Olga García-Obregón (2), Sur Herrera-Paredes (1)
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Abstract:
Systemic lupus erythematosus (SLE) is an autoimmune disease characterized by chronic inflammation, multi-organ damage, and a relapsing–remitting clinical course. Environmental factors play a significant role in triggering lupus flares and include hormones, medications, ultraviolet radiation, infections, toxins, cigarette smoking, as well as dietary factors and the gut microbiome, all of which contribute to immune activation and disease exacerbation. Patients with SLE exhibit alterations in gut microbiome composition, commonly referred to as intestinal dysbiosis, which has been associated with increased intestinal permeability, facilitating the translocation of bacterial components that may trigger systemic inflammation and autoimmune responses, thereby

contributing to disease flares and organ damage. However, current research provides limited information on differences in gut microbiome composition between patients with SLE and healthy controls in relation to diet quality. By examining this interaction, we aim to improve understanding of the role of environmental factors in SLE aetiology and support the development of novel therapeutic strategies by evaluating differences in gut microbiome composition between patients with SLE and healthy participants while simultaneously assessing diet quality. We are initiating this project in collaboration with the Registro Mexicano de Lupus (LupusRGMX), and I will present a preliminary descriptive analysis of survey data on diet quality in patients with SLE. Through this analysis, we examined associations between diet quality and variables such as age, comorbidity incidence, and cohabitation status. This preliminary work will provide a broader overview of current dietary habits in the SLE population and guide the design and hypotheses of the main study. We aim for this study to establish a foundation for future research focused on exploring causal relationships between gut microbiome dysbiosis and SLE development and on evaluating gut microbiome-based biomarkers supporting the potential role of dietary intervention for disease prevention and management with possible effects on symptomatology and disease progression.

Identifying ecological interactions mediated by biosynthetic gene clusters in the ocean microbiome

Presenter: Andrea Zermeño Díaz
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Abstract:
The diversity of microbial communities in the ocean is immense; and the ecological interactions among microorganisms further enhance it. These interactions can occur through secondary metabolites produced by biosynthetic gene clusters (BGCs), many of which are of biomedical and biotechnological interest, such as antibiotics and pesticides. Despite their relevance, our understanding of their impact on ecological interactions remains limited. In this study, we identified potential molecular inter-lineage interactions among marine microorganisms (bacteria and archaea) using 26,293 metagenome-assembled genomes and 28,109 predicted biosynthetic gene clusters from the Ocean Microbiomics Database v1.1. We characterized patterns of co-occurrence and co-exclusion between microbial lineages and BGCs, grouping BGCs into families and clans based on overall biosynthetic and sequence similarity. The statistical significance of these associations was evaluated using a binomial test, and we systematically assessed how different levels of taxonomic and biosynthetic resolution affected the strength of the statistical signal. Across all resolutions, co-occurrence signals were consistently stronger than co-exclusion signals, with vastly fewer significant co-exclusion than co-occurrence associations. We represented these associations as co-occurrence and co-exclusion networks, revealing bacteriocins as the major contributors to the interactions. In the future, we aim to explore changes in these patterns across global environmental gradients such as depth, oxygen levels and temperature, which will provide insights into how environmental pressures shape microbial community structure. This study will produce specific hypotheses regarding the molecular basis of ecological dynamics among marine microorganisms and provide a broader understanding of the contribution of BGCs in microbial ecosystems.

Computational Population Genetics Lab

Responsable: Dr. Diego Ortega Del Vecchyo

The impact of directional selection on patterns of genetic and phenotypic variation on a continuous space

Presenter: Valeria Cabrera Rojas
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Abstract:
The relationship between the strength of directional selection and the phenotype of individuals in an environment is a widely studied topic In Evolutionary Biology. Its study allows us to anticipate how populations respond to selective pressure such as climate changes, the availability of different resources and the competition with different species. However, the majority of theoretical models do not take into account a geographical context which can also shape both patterns of phenotypic and genetic variation. We need to take the geographical position of individuals into account to study directional selection to understand the dynamics of this process in spatial models with changing and heterogeneous environments. In this study we perform simulations to study how environmental factors that vary over space and directional selection can impact phenotypic values and QTLs that have an effect on a particular phenotype. We hope to devise general rules on how gene flow, changing environmental factors and different strengths of directional selection shape the evolution of phenotypes and the genetic variants that have an impact on complex traits.

Reconstrucción Paleogenómica del Desierto Chihuahuense: Explorando la Historia Evolutiva a través del sedaDNA

Presenter: Laura Figueroa Corona
Authors: Laura Figueroa-Corona (1), Alejandra Castillo (1), Vania Ferrer Parra (2), Barbara Moguel (3), Santiago Rosas-Plaza (1), Mariano Cerca Martínez (2), Federico Sanchez Quinto (1), Diego Ortega Del Vecchyo (1) , Marcela Sandoval Velasco (4)
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3: Departamento de Ciencias Químico - Biológicas en la Universidad de las Américas, Puebla, Puebla, México.
4: Centro de Ciencias Genómicas (CCG), Universidad Nacional Autónoma de México (UNAM), Campus Morelos„ México.

Abstract:
El Desierto Chihuahuense es una de las ecorregiones más biodiversas de América del Norte y su paisaje actual es el resultado de transformaciones ambientales ocurridas en el pasado. Durante el último periodo glacial, este desierto fue un corredor boscoso compuesto por pinos piñoneros, enebros y robles. Gradualmente esta región se transformó en un ecosistema árido a partir de la reducción de paleolagos formados en los ciclos glaciares. De los cuerpos de agua más importantes y extensos de Norteamérica es el paleolago Irritila que ocupó una extensión aproximada de 15,000 km² y albergó una gran diversidad de moluscos durante el Cuaternario tardío. Este proyecto busca reconstruir la diversidad que habitaba en esta región durante el Pleistoceno a partir de muestras sedimentarias y la recuperación de ADN antiguo (sedaADN) recolectado en los márgenes del paleolago de Irritila en Viesca, Coahuila. Se colectó un total de 70 muestras sedimentarias de 6 perfiles. Los resultados preliminares de los perfiles geológicos y las dataciones por radiocarbono a partir de 14 muestras de sedimento confirman una temporalidad de 670 a 9020 años. Asimismo, se realizaron las identificaciones taxonómicas de moluscos, encontrándose dos especies endémicas, Spinopyrgus luismaedai y Pyrgulopsis paleominckleyi.

Hasta este momento se han generado un total de 2498.8 millones de reads de 5 de los sitios muestreados, los cuales analizaremos utilizando una aproximación metagenómica. En los que evaluaremos la proporción de secuencias vegetales y de moluscos presentes en cada una de las muestras, buscando reconstruir la diversidad genética de especies mediante el análisis de genes altamente conservados. Con esto, se evaluará la distribución histórica y conectividad de las especies que habitan y habitaron en El Desierto Chihuahuense. Además, se reconstruirán los escenarios histórico-demográficos que expliquen los cambios a lo largo del tiempo.

Analysis of a Possible Relaxation of Natural Selection in Human Populations

Presenter: Miriam Alejandra Jimenez Marmolejo
Authors: M. Alejandra Jimenez Marmolejo (1), Diego Ortega-Del Vecchyo (1)
Affiliations:
1: Laboratorio Internacional de Investigación sobre el Genoma Humano (LIIGH-UNAM).

Abstract:
Purifying selection is an evolutionary force that removes deleterious genetic variants from populations. The efficacy of purifying selection in humans has been hypothesized to have changed recently on certain genes coinciding with environmental and cultural transitions including the development of agriculture and modern medicine. These transitions are proposed to have modified selective pressures by changing the fitness effects of deleterious variants resulting in a relaxation of purifying selection, whereby mutations that were deleterious in the past are less deleterious in the present. Here, we investigate how the relaxation of purifying selection changes genetic variation patterns. We used forward-time simulations varying the relaxation of purifying selection acting on deleterious mutations. The resulting genetic variation patterns were analyzed using the site frequency spectrum (SFS) by comparing variant counts across distinct frequency classes. The relaxation of purifying selection resulted in a consistent enrichment of rare variants within a derived allele frequency window of 0.75–1.50%, while very low (<0.5%), intermediate, and high-frequency classes remained largely unchanged. We show plans to extend this analysis by integrating genealogical information and analyzing different demographic scenarios consistent with human history with the goal of evaluating evidence for the relaxation of purifying selection in humans.

Distribución de los pósters

Sesión de Pósters 1 & Coffee Break

Número	Ponente	Título	Laboratorio
1	Laura Carrillo Olivas	Tracing Introduced Pathogen Lineages and Social Dynamics in Colonial Mexico City through Paleogenomics	Population and Evolutionary Genomics Lab (Dra. María C. Ávila Arcos)
2	David Figueroa	Genomic Insights into Functional Adaptation of Non-Model Yeasts in Traditional Agave Fermentations in Mexico	Genome Evolution Lab (Dra. Lucía Morales)
3	Miguel Ángel Flores Varela	Ancient virus identification through ancient DNA in prehispanic and colonial human remains in Mexico	Population and Evolutionary Genomics Lab (Dra. María C. Ávila Arcos)
4	María Fernanda García Rodríguez	Genomic history and ancestral origins of Afro-Mexican populations	Population and Evolutionary Genomics Lab (Dra. María C. Ávila Arcos)
5	Valeria Gómez-Vela	Genomic findings and SHANK 3 microduplication in a patient with autism spectrum disorder	Mendelian Genomics and Precision Health Lab (Dra. Claudia Gonzaga Jauregui)
6	Mariana G. Guerrero Osornio	Temporal Dynamics of Yeasts, Bacteria and Viruses During Agave Fermentations	Genome Evolution Lab (Dra. Lucía Morales)
7	Ernesto Gutiérrez Piñón	Effects of initial strain proportions on competitive dynamics in *Bacillus cocultures* from Cuatro Ciénegas	Ecology and Evolution Lab (Dr. Sur Herrera Paredes)
8	Etzael Alexis Mendoza García	Early Metabolic Effects of High-Fat Diets in a Mouse Model of MASLD	Cancer Genetics and Bioinformatics Lab (Dra. Daniela Robles)
9	Abigail Montante Arenas	The impact of metaboliC exchange on bacterial community dynamics	Ecology and Evolution Lab (Dr. Sur Herrera Paredes)
10	Alejandro Pinto Sánchez	Detecting *Physarum polycephalum* decision making using an object detection model	Ecology and Evolution Lab (Dr. Sur Herrera Paredes)
11	Maria Fernanda Requena Romo	A MetaHiC-Based Pipeline for Detecting Yeast Hybrids	Genome Evolution Lab (Dra. Lucía Morales)
12	Mónica Reyes Ramírez	Comparative genomic of cellular complexity in Thiotrichales	Ecology and Evolution Lab (Dr. Sur Herrera Paredes)
13	Giovanni Emmanuel Rodríguez González	Impact of different high-fat diet intakes and MASLD development in mouse hepatic health and zonation	Cancer Genetics and Bioinformatics Lab (Dra. Daniela Robles)
14	Victoria Rodríguez Lelis	Mathematical modeling of negative feedback in maintenance of telomere homeostasis	Evolutionary Systems Biology Lab (Dra. Mariana Gómez Schiavón)
15	Cielo del Carmen Rodríguez Molina	Factores clínicos asociados a nefritis lúpica en población mexicana	Regulatory Genomics and Bioinformatics Lab (Dra. Alejandra Medina Rivera)
16	Edgar Pavel Salazar Fernández	Analysis of a deeply-phenotyped familial hypercholesterolemia cohort from Mexico shows a role for both rare and common alleles across known dyslipidemia genes and reveals structural variation in a novel locus	Paleogenomics and Evolutionary Biology Lab (Dr. Federico Sánchez Quinto)
17	Iván Sedeño	Bidirectional gene flow in populations of *S. paradoxus* and *S. cerevisiae* from agave fermentations	Genome Evolution Lab (Dra. Lucía Morales)
18	Zyanya Valentina Velazquez Aldrete	Pulmonary Fibrosis: A failure of homeostatic control	Evolutionary Systems Biology Lab (Dra. Mariana Gómez Schiavón)

Sesión de Pósters 2 & Coffee Break

Número	Ponente	Título	Laboratorio
19	Diana Barrientos González	Between Growth and Invasion: Regulatory Dynamics of Melanoma Phenotypes	Evolutionary Systems Biology Lab (Dra. Mariana Gómez Schiavón)
20	Román Cervantes Levario	Identification of disease-associated variants in Heme production genes associated with Porphyrias	Mendelian Genomics and Precision Health Lab (Dra. Claudia Gonzaga Jauregui)
21	Leonardo Yair Correa Mendoza	Análisis de las afinidades genéticas y estructura social de los individuos prehispánicos excavados en Tlatel 7 de Santa Lucía I	Paleogenomics and Evolutionary Biology Lab (Dr. Federico Sánchez Quinto)
22	Salvador Alejandro Cuevas Villicaña	Identification and validation of therapeutic targets for acral melanoma	Cancer Genetics and Bioinformatics Lab (Dra. Daniela Robles)
23	Diana Delgado Gutiérrez	Analysis of Genetic Variants Associated with Parkinson's Disease and Depression	Regulatory Genomics and Bioinformatics Lab (Dra. Alejandra Medina Rivera)
24	Mariana Fraustro Méndez	Cognitive Impairment in Parkinson's Disease: Association between White Matter Lesion Volume and MoCA Score	Regulatory Genomics and Bioinformatics Lab (Dra. Alejandra Medina Rivera)
25	Kitzia Gómez Cepeda	Modelado funcional en *Caenorhabditis elegans* de variantes genéticas en el complejo PP2A asociadas al Síndrome de Houge-Janssens	Mendelian Genomics and Precision Health Lab (Dra. Claudia Gonzaga Jauregui)
26	Yael Daniel Hernandez Gonzalez	Plasticity Is Not Just "Change": Formalizing the Concept Beyond Semantic Ambiguity	Evolutionary Systems Biology Lab (Dra. Mariana Gómez Schiavón)
27	Emmanuel Hernández Sánchez	Generating phenotypic heterogeneity in fluctuating environments: Between Oscillations and Bistability	Evolutionary Systems Biology Lab (Dra. Mariana Gómez Schiavón)
28	Dianella Iglesias Rodríguez	Whole slide image analysis of mexican patients with melanoma acral using QuPath	Cancer Genetics and Bioinformatics Lab (Dra. Daniela Robles)
29	Diego Alejandro Marquez Cerna	Genomic and Transcriptomic Characterization of a *Saccharomyces cerevisiae* × *Saccharomyces paradoxus* Hybrid Yeast Strain	Genome Evolution Lab (Dra. Lucía Morales)
30	Diego Morales Martínez	Bistability and Transcriptional Bursting: A Comparative Study of Noise-Driven Switching Dynamics	Evolutionary Systems Biology Lab (Dra. Mariana Gómez Schiavón)
31	Rigoberto Padilla Bustos	Molecular sexing of ancient faunal remains: methods, limits and prospects	Paleogenomics and Evolutionary Biology Lab (Dr. Federico Sánchez Quinto)
32	Natalie Berenice Pineda Morán	Identificación taxonómica de murciélago desconocido utilizando datos genómicos	Paleogenomics and Evolutionary Biology Lab (Dr. Federico Sánchez Quinto)
33	Ariana Gabriela Reyes Silva	In vivo functional modeling of SMARCA5 variants associated with a novel neurodevelopmental disorder in *Caenorhabditis elegans*	Mendelian Genomics and Precision Health Lab (Dra. Claudia Gonzaga Jauregui)
34	Julieta Berenice Rivera Zavala	Disruption of energy balance and metabolic rhythmicity induced by a High-Fat Diet in MASLD development	Cancer Genetics and Bioinformatics Lab (Dra. Daniela Robles)
35	Erandi Abril Salas Romero	Panorama actual del tamizaje neonatal genómico para la detección temprana de enfermedades genéticas en el mundo	Mendelian Genomics and Precision Health Lab (Dra. Claudia Gonzaga Jauregui)
36	María Fernanda Talavera Cruz	Mechanical Stress as a Contributing Factor in Acral Melanoma	Cancer Genetics and Bioinformatics Lab (Dra. Daniela Robles)
37	Karen Astrid Zárate Saldierna	Influencia de los factores reproductivos en la edad de inicio de la enfermedad de Párkinson en mujeres mexicanas	Regulatory Genomics and Bioinformatics Lab (Dra. Alejandra Medina Rivera)

Resumen de las sesiones de pósters

Cancer Genetics and Bioinformatics Lab

Responsable: Dra. Daniela Robles

Poster 8: Early Metabolic Effects of High-Fat Diets in a Mouse Model of MASLD
Early Metabolic Effects of High-Fat Diets in a Mouse Model of MASLD
Presenter: Etzael Alexis Mendoza García
Authors: Mendoza-García Etzael Alexis (1), Gutiérrez-García Claudia (1), Rivera-Zavala Julieta Berenice (1), Rodríguez-González Giovanni Emmanuel (1), Rochefort-García Ana Paola (1), Boulogne-Seda Nelson Alejandro (1), Adams David J (7), Ledesma-Juárez Osiel (3), Bastidas-Ponce Aimée (4), Díaz-Muñoz Mauricio (5), Miller Derek (6), Strathdee Douglas (6), Robles-Espinoza Carla Daniela (1), Molina-Aguilar Christian (1)
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Abstract:
In Mexico, 36.9% of the adult population suffers from obesity, and this figure is projected to reach 45% by 2030. Among the main factors contributing to the development of this condition are poor-quality diets and the consumption of high-fat foods, as well as sedentary lifestyles. These factors promote the onset of metabolic diseases such as metabolic dysfunction–associated steatotic liver disease (MASLD), which affects more than 30% of the global population and represents a growing public health concern. MASLD encompasses a spectrum of hepatic alterations ranging from simple steatosis to metabolic dysfunction–associated steatohepatitis (MASH), which can progress to fibrosis, cirrhosis, and even hepatocellular carcinoma. Obesity is one of the major risk factors for MASLD, with disruption of lipid metabolism being a central component of its pathophysiology. Hepatic energy imbalance occurs when triglyceride accumulation exceeds the liver's capacity for metabolism or export in the form of lipoproteins. This process is further exacerbated by high fructose intake, which stimulates hepatic de novo lipogenesis and promotes inflammatory states. MASLD is often a subclinical condition, which complicates early diagnosis and allows for silent disease progression. In this study, we evaluated the impact of high-fat diets on the early development of metabolic alterations associated with MASLD in C57BL/6J mice. Animals were fed a control diet (CHOW) or high-fat diets containing 10% or 40% fat. Serum biochemical analyses and anthropometric measurements were performed at different time points. Our results show that the 40% fat diet induces significant metabolic alterations after 16 weeks, evidenced by increased total cholesterol levels and impaired glucose regulation, whereas the 10% fat diet produces moderate changes consistent with partial metabolic adaptation. These findings indicate that the metabolic impact of diet depends on fat content and duration of exposure, even at early stages prior to the establishment of advanced liver damage.

Poster 13: Impact of different high-fat diet intakes and MASLD development in mouse hepatic health and zonation.
Impact of different high-fat diet intakes and MASLD development in mouse hepatic health and zonation.
Presenter: Giovanni Emmanuel Rodríguez González
Authors: Rodríguez-González Giovanni Emmanuel (1), Rivera-Zavala Julieta Berenice (1),

Gutiérrez-García Claudia (1), Adams David J (7), Mendoza-García Etzael Alexis (1), Rochefort-García Ana Paola (1), Boulogne-Seda Nelson Alejandro (1), Martínez-García Zuly Dannae (2), Ledesma-Juárez Osiel (3), Bastidas-Ponce Aimée (4), Díaz-Muñoz Mauricio (5), Miller Derek (6), Strathdee Douglas (6), Robles-Espinoza Carla Daniela (1), Molina-Aguilar Christian (1)
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Abstract:
Metabolic dysfunction-associated steatotic liver disease (MASLD) is a chronic liver disease affecting 35% of the population, recognized as the leading cause of liver-related morbidity and mortality. It is characterized by an hepatic fat infiltration that surpasses 5% of total liver mass, and is related to factors such as obesity, hypertension, and insulin resistance. Periportal-to-pericentral reprogramming triggered in MASLD by sustained steatosis, hypoxia, oxidative stress and other alterations, causes a loss of identity in hepatocytes (HEPs) and thus changes typical expression of zonation genes. Moreover, several transcriptional changes in HEPs have been linked as response to high-fat diets (HFDs); for example, promotion of lipid droplets (LDs) associated with lipid synthesis upregulation. Methods: Five-week-old male C57BL/6 mice were fed daily with a chow diet, a 10% fat diet, or a 40% fat diet for 0 weeks (healthy liver), 16 weeks (MASLD), and 34 weeks (fibrotic liver). Body weight and food intake were monitored throughout the experimental period. After each timeframe, subjects were sacrificed previous to liver extraction. Histologic analysis of liver samples was conducted using hematoxylin and eosin (HandE) staining and immunofluorescence (IF) assays aimed at glutamine synthetase expression. Results: IF assay evaluation of healthy liver samples showed signs of adaptation after a 4 day intake of the 10% fat diet as a change in zonation. Besides, in samples corresponding to the 16 week timeframe, clear signs of steatosis were found in the 40% fat diet, while those corresponding to the 10% fat intake experienced an increase in LDs. Conclusion: Cellular adaptation to HFDs can be found as early as the first week of intake. Furthermore, after constant exposure to these diets, signs of transcriptomic modifications of HEPs can be found. The adaptation of glutamine synthetase marker may also suggest a disruption in the β-catenin-dependent zonation.

Poster 22: Identification and validation of therapeutic targets for acral melanoma
Identification and validation of therapeutic targets for acral melanoma

Presenter: Salvador Alejandro Cuevas Villicaña
Authors: Salvador Alejandro Cuevas-Villicaña (1), Christian Molina Aguilar (1), Dr. Rodrigo González Barrios de la Parra (2), Mauricio Diaz Muñoz (3), Dr. Floris Foijer (4), Dr. Patricia Abrão Possik (5); Dr.Carla Daniela Robles Espinoza (1)
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4: European Research Institute for the Biology of Ageing (ERIBA-UG).
5: Instituto Nacional de Cancer Brasil (INCA).

Abstract:
Acral melanoma (AM) represents a public health problem, particularly as it is the most prevalent and aggressive melanoma subtype in Latin American, African, and Asian populations. Unlike other melanomas, its pathogenesis is not determined by UV radiation but is instead characterized by high chromosomal instability (CI) and a prevalence of structural variants in the genome.

This biology contributes to its poor prognosis and its often-suboptimal response to conventional immunotherapies and targeted agents, which creates a need for new therapeutic strategies beyond traditional chemotherapy. This project addresses this therapeutic gap by studying the genomic characteristics of AM. It is hypothesized that high CI activates the cGAS-STING-IL-6 pro-survival inflammatory signaling pathway, making tumors dependent on the interleukin-6 receptor (IL-6R). The key contribution is the proposal to repurpose Tocilizumab, an IL-6R inhibitor clinically approved to treat rheumatoid arthritis and that, according to previous studies conducted at the University of Groningen, is a possible treatment alternative for cancers with high CI and IL-6R overexpression. This project seeks to validate CI and IL-6R overexpression as predictive biomarkers in AM, offering a rational and personalized treatment strategy. The main beneficiaries of this research are AM patients, especially those in underrepresented populations such as Mexico and Brazil, who currently face limited options and worse prognoses. The translational application of this research is substantial. One result will be the generation of the first patient-derived xenograft (PDX) models of AM from Latin American patients, which will help reduce the gap in preclinical research tools. These models are important for personalized medicine, allowing for trials that can present treatment alternatives and, eventually, the clinical adoption of Tocilizumab as a possible immunotherapy for this disease.

Poster 28: Whole slide image analysis of mexican patients with melanoma acral using QuPath

Whole slide image analysis of mexican patients with melanoma acral using QuPath

Presenter: Dianella Iglesias Rodríguez
Authors: Dianella Iglesias (1), Lizbett Hidalgo Pérez (2), David J. Adams (3), Mark Tullett (4), Mark J. Arends (4), Carla Daniela Robles-Espinoza (1,3)
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4: Edinburgh Pathology, Cancer Research UK Scotland Centre, Institute of Genetics and Cancer, University of Edinburgh, Edinburgh EH4 2XU, UK.

Abstract:
Acral melanoma is the most common type of melanoma in Mexico. Early diagnosis is crucial, whereby the implementation of highly sensitive methods based on non-invasive strategies is important to improve disease prognosis, and to increase the specificity of evaluation criteria. Histopathology analysis supported by algorithms of Artificial Intelligence has been hypothesised to be more accurate than histopathology evaluation on its own. The aim of this project is to analyze whole slide images of tumours from Mexican patients with acral melanoma using QuPath. A study was conducted in collaboration with physicians and oncologists from INCan, Mexico City, where a paraffin-embedded tumor sample was taken from patients diagnosed with acral melanoma. Tumor samples were prepared on slides and stained with hematoxylin/eosin. High-resolution images were taken from each tumor at the University of Edinburgh, Scotland. images were analyzed using the QuPath program v (0.6.0). A color deconvolution was applied using the Estimate Stain vector command. An annotation of the image analysis was performed using the positive cell detection command, which will help to determine the total number of positive cells within the annotated area. It used the number of positive cells and the detected area to calculate the average number of positive cells per um². The correlation between the number of positive cells and the stage of the disease was calculated. Usually, the correlation between the number of positive cells and the stage of the disease was positive. The average number of positive cells per um² didn't always coincide with the highest number of tumor cells calculated by imaging. In several images, cells related to inflammation were found independently of the stage of disease. The results of these histological studies in conjunction with transcriptomic or genomic results could help to impute or predict DNA damage in cancer research.

Poster 34: Disruption of energy balance and metabolic rhythmicity induced by a High-Fat Diet in MASLD development

Disruption of energy balance and metabolic rhythmicity induced by a High-Fat Diet in MASLD development

Presenter: Julieta Berenice Rivera Zavala

Authors: Rivera-Zavala Julieta Berenice(1), Gutiérrez-García Claudia(1), Mendoza-García Etzael Alexis(1), González Giovanni Emmanuel(1), Rochefort-García Ana Paola(1), Boulogne-Seda Nelson Alejandro(1), Ledesma-Juárez Osiel(2), Díaz-Muñoz Mauricio(3), Miller Derek(4), Strathdee Douglas(4), Adams David J(5), Molina-Aguilar Christian(1) and Robles-Espinoza Carla Daniela(1)

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5: The Wellcome Trust Sanger Institute.

Abstract:

Metabolic dysfunction-associated steatotic liver disease (MASLD) is a chronic liver disease and a leading cause of liver-related mortality. Factors associated with MASLD include obesity, type 2 diabetes, dyslipidemia, lifestyle factors, and genetic predisposition. MASLD is driven by metabolic dysfunction, including insulin resistance, increased hepatic lipid influx, de novo lipogenesis, and impaired lipid oxidation, leading to hepatic steatosis (>5% fat accumulation). Lipotoxicity from accumulated lipids can promote inflammation, fibrosis, and progression to hepatocellular carcinoma (HCC). A microsimulation model estimated that MASLD prevalence will increase from 33.7% in 2020 to 41.4% by 2050, representing a significant public health burden worldwide. Methods: Five-week-old male C57BL/6 mice were fed daily with a chow diet, a 10% fat diet, or a 40% fat diet for 0 weeks (healthy liver), 16 weeks (MASLD), and 34 weeks (fibrotic liver). Body weight and food intake were monitored throughout the experimental period. During the final six days, animals were individually housed in metabolic cages to measure oxygen consumption (VO□) and carbon dioxide production (VCO□). These measurements were used to calculate the respiratory quotient (RQ) as an indicator of substrate utilization: carbohydrates (1.0), proteins (0.8–0.9), and lipids (0.7–0.8). Total energy expenditure and locomotor activity (horizontal and vertical) were assessed continuously over 24 hours. Results: All evaluated parameters showed loss of circadian rhythmicity in mice fed 10% and 40% fat diets. RQ values indicated increased lipid utilization throughout the day, while total energy expenditure decreased over the 24-hour period. Conclusion: Chronic high-fat feeding (16 weeks) disrupts metabolic rhythmicity and alters substrate utilization. Sustained lipid oxidation may increase mitochondrial stress and induce a lipotoxic state, which can trigger altered immune responses and activate cells involved in hepatic fibrosis. In the context of obesity, these changes are associated with reduced energy expenditure and locomotor activity.

Poster 36: Mechanical Stress as a Contributing Factor in Acral Melanoma

Mechanical Stress as a Contributing Factor in Acral Melanoma

Presenter: María Fernanda Talavera Cruz

Authors: M. Fernanda Talavera-Cruz (1), Johan Rott (2), Rotem Leshem (2), C. Daniela Robles-Espinoza (1,3), Kerrie Marie (2)

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3: Wellcome Sanger Institute, Hinxton, UK.

Abstract:

Acral melanoma is a rare subtype of melanoma that arises on the palms and soles and shows a

higher prevalence in Mexico compared to many other countries. Unlike cutaneous melanomas, acral melanoma is not associated with ultraviolet radiation exposure. Its frequent occurrence in pressure-bearing areas suggests that mechanical forces may play a role in disease development. This project aimed to investigate the potential contribution of mechanotransduction pathways to acral melanoma biology. To test this hypothesis, a differential gene expression analysis was performed on RNA-sequencing data obtained from acral melanoma patient samples. This analysis identified several candidate genes with putative roles in mechanosensing regulation. Selected candidates were subsequently examined in vitro using melanoma cell culture models combined with a mechanosensitive stretching system. The effects of mechanical stimulation on gene expression, cellular morphology, and behaviour were assessed using immunofluorescence staining and confocal microscopy. Preliminary findings indicate that mechanical stretching induces notable morphological changes in melanoma cells, particularly affecting the formation and appearance of filopodia. Filopodia are actin-rich plasma membrane protrusions that function as cellular antennae, enabling cells to sense and respond to their microenvironment. These structures are critically involved in processes such as cell migration, wound healing, adhesion to the extracellular matrix, guidance toward chemoattractants, neuronal growth-cone pathfinding, and embryonic development. Together, these results support a potential role for mechanical forces in influencing acral melanoma cell behaviour and suggest that altered mechanosensing and cytoskeletal dynamics may contribute to disease pathogenesis. Further investigation of these pathways may provide new insights into the unique biology of acral melanoma.

Ecology and Evolution Lab

Responsable: Dr. Sur Herrera Paredes

Poster 7: Effects of initial strain proportions on competitive dynamics in Bacillus cocultures from Cuatro Ciénegas

Effects of initial strain proportions on competitive dynamics in Bacillus cocultures from Cuatro Ciénegas

Presenter: Ernesto Gutiérrez Piñón
Authors: Ernesto Gutiérrez Piñón[1,2], Natalia Said Muñoz[1], Sur Herrera Paredes[1]
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Abstract:
Cuatro Ciénegas, Coahuila, is an oligotrophic oasis that harbors high microbial diversity and serves as a model system for the study of microbial ecology. In this work, we evaluated competitive interactions between two Bacillus strains (Bacillus sp. CH111 and Bacillus sp. CH450) isolated from this ecosystem, with the aim of assessing how initial strain proportions influence competitive outcomes in coculture. The strains were grown separately, equalized by optical density (OD600), and mixed to form synthetic communities at different initial proportions (1:1, 1:2, and 2:1). Competitive dynamics were evaluated by quantifying colony-forming units (CFU/mL), including a replicate in which cocultures were maintained and monitored over three consecutive days. Preliminary results suggest that Bacillus sp. CH450 maintains higher and more stable abundances across proportions, indicating a potential competitive advantage over Bacillus sp. CH111, whose abundance was more strongly affected by initial conditions. Additionally, this study highlights limitations in the use of optical density as a proxy for bacterial abundance in competitive systems, motivating future work focused on the calibration and implementation of complementary quantitative approaches.

Poster 9: The impact of metaboliC exchange on bacterial community dynamics
The impact of metaboliC exchange on bacterial community dynamics
Presenter: Abigail Montante Arenas
Authors: Abigail Montante Arenas(1,2), Natalia Said Muñoz(1,3), Sur Herrera Paredes(1)
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Abstract:
The rhizosphere is a hotspot for bacterial communities. These communities exhibit a dynamic composition in which the abundance and diversity of their members change over time. Within a community, there exists an extensive network of metabolic chemical reactions and metabolic exchange, which we define as syntrophy, where a metabolic product of an organism is subsequently consumed as a substrate by other organisms. Using a set of ten rhizospheric bacterial strains, we simulated in silico bacterial communities with the Computation of Microbial Ecosystems in Time and Space (COMETS) framework, which is based on Dynamic Flux Balance Analysis (dFBA). In this approach, the abundance of different species and environmental metabolites are treated as dynamic variables, enabling the simulation of the growth and interactions of bacterial communities within a spatially defined environment. We have observed that individual bacterial growth properties alter within a community, where dynamics depend on the specific members present. Our objective is to identify how metabolic resource exchange shapes the structure and composition of microbial communities and contributes to the emergence of ecological relationships.

Poster 10: Detecting Physarum Polycephalum decision making using an object detection model
Detecting Physarum Polycephalum decision making using an object detection model
Presenter: Alejandro Pinto Sánchez
Authors: Alejandro Pinto Sánchez (1, 2), Rodrigo Aguilar Díaz (1, 2), Onna Nayyu Leyva Alcántara (1, 2), Sur Herrera Paredes (2)
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2: Laboratorio Internacional de Investigación sobre el Genoma Humano (LIIGH-UNAM).

Abstract:
Physarum polycephalum is a multinucleate slime mold that is commonly cultivated in lab settings. This organism uses its body, called plasmodium, to receive chemical and physical cues from its environment to perform computations similar to what we call cognition. Among other interesting features, P. polycephalum is able to decide the best route to get to food (i.e. shortest path problem), to decide between two paths and choose whichever has the most food or to remember the most likely route to get to food, all of this without having any kind of nervous system. In this study, we're using a YOLO-based object detection model that has been trained with images of previous experiments to quantify features of P. polycephalum. Some of the previous research has used a qualitative analysis of video footage or images to describe the behavior of P. polycephalum, however, this comes with two main challenges. First, it's challenging to identify significant patterns across individuals, such as decision-making latency or velocity. Second, manual analysis becomes more labor-intensive and unreliable when working with larger datasets.The aim of the model we made is (a) to objectively measure behavioral parameters such as movement direction, velocity, and surface area, and (b) to standardize the process of analysis so that it is easier to work with larger volumes of samples. This analysis can help us understand more robustly how this organism optimizes its decision making strategies in different experimental contexts.

Poster 12: Comparative genomic of cellular complexity in Thiotrichales
Comparative genomic of cellular complexity in Thiotrichales

Presenter: Mónica Reyes Ramírez
Authors: Monica Reyes Ramírez[1,2], Andrea Zermeño Díaz[1,2], Sur Herrera Paredes[1]
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2: Licenciatura en Ciencias Genómicas, Juriquilla, UNAM.

Abstract:
Thiotrichales are filamentous gammaproteobacteria, that predominantly inhabit anoxic marine sediments and obtain energy through the oxidation of sulfur compounds. Within this order, bacteria with exceptionally large genomes have been described, such as Candidatus Thiomargarita magnifica, which is characterized by a unique morphology and a genome size far exceeding the bacterial average. These features position it as an extreme case and a reference point for exploring genomic diversity and complexity within the Thiotrichales. This study aims to characterize cellular complexity and biosynthetic potential across Thiotrichales species, and to evaluate whether Ca. T. magnifica represents an extreme case within a broader trend across the order. Cellular complexity was assessed using genome size, the number of coding sequences (CDSs), and the abundance of genes associated with cell elongation and division. Biosynthetic potential was quantified based on the genomic extent of biosynthetic gene clusters (BGCs) and the proportion of the genome dedicated to secondary metabolism. A gradient in biosynthetic capacity was identified, with higher values in the genus Thiomargarita compared to other genera within the order, and a pronounced extreme in Ca. T. magnifica, suggesting progressive adaptations associated with competitive advantages conferred by secondary metabolite production in anoxic sedimentary environments

Evolutionary Systems Biology Lab

Responsable: Dra. Mariana Gómez Schiavón

Poster 14: Mathematical modeling of negative feedback in maintenance of telomere homeostasis
Mathematical modeling of negative feedback in maintenance of telomere homeostasis
Presenter: Victoria Rodríguez Lelis
Authors: Victoria Rodríguez Lelis [1, 2], Mariana Gómez Schiavón [1, 3]
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3: Millennium Science Initiative Program, Millennium Institute for Integrative Biology (iBio), Chilean National Agency for Research and Development, Santiago, Chile.

Abstract:
Telomeres are repetitive DNA sequences located at the ends of chromosomes, essential for protecting genomic integrity. Maintaining an optimal telomere length is crucial for this function. Critically short telomeres trigger DNA damage responses and genomic instability. Conversely, excessively long telomeres—often sustained by persistent telomerase activity—are associated with cellular immortalization and cancer. The regulation of telomere length involves a dynamic interplay between the telomerase enzyme and the shelterin protein complex. Here we propose that a negative feedback loop, mediated by shelterin complexes and telomerase, constitutes the core mechanism for sustaining telomere length homeostasis. To explore this hypothesis, we developed a simple mathematical model based on a regulatory mechanism where the number of shelterin complexes bound to a telomere is length-dependent. In this model, long telomeres accumulate more shelterin complexes, which strongly inhibits telomerase binding and prevents telomere elongation. Conversely, short telomeres, with fewer bound shelterin complexes,

present minimal inhibition, permitting telomerase access and subsequent elongation. Through dynamical analysis of the model, we aim to characterize the system's homeostatic properties and evaluate how genetic perturbations (e.g., mutations in shelterin complex or telomerase) can disrupt stability. This work provides a theoretical framework to explore how negative feedback governs homeostatic capacity in telomere length regulation, thereby elucidating its robustness and failure modes.

Poster 18: PULMONARY FIBROSIS: A FAILURE OF HOMEOSTATIC CONTROL
PULMONARY FIBROSIS: A FAILURE OF HOMEOSTATIC CONTROL
Presenter: Zyanya Valentina Velazquez Aldrete
Authors: Zyanya Valentina Velazquez Aldrete(1), Mariana Gómez-Schiavon(2,3)
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Abstract:
Pulmonary fibrosis is a chronic inflammatory disease characterized by a dysregulated and perpetually active repair process, leading to the loss of immune-regulatory homeostasis and the establishment of a new pathological "steady state." Tissue damage in the alveolar epithelium triggers a cascade of highly conserved and strictly regulated repair responses that, under normal conditions, resolve in an orderly manner. However, in pulmonary fibrosis, this process is disrupted. Currently, there is no cure for this disease, but there is a great effort to understand the pathways of immune response modulation, tissue repair, and therapeutic strategies to inhibit fibrotic progression. In this work, we present an approach aimed at identifying the main key regulatory pathways to evaluate the homeostatic capacity of the system and recognize stimuli and responses that determine the transition of the disease from healthy state to a fibrotic one after an initial injury. We are developing a minimal mathematical mechanistic model to evaluate the contributions of key components that determine the stability of the system and its vulnerability, and to determine its susceptibility to fibrotic transformation. This approach will allow us to quantitatively evaluate the homeostatic capacity of the tissue, and recognize the critical stimuli that trigger the pathological transition.

Poster 19: Between Growth and Invasion: Regulatory Dynamics of Melanoma Phenotypes
Between Growth and Invasion: Regulatory Dynamics of Melanoma Phenotypes
Presenter: Diana Barrientos González
Authors: Diana Barrientos González (1), Isabel Montejano Montelongo (1), Mariana Gómez Schiavon(1,2)
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2: Millennium Science Initiative Program, Millennium Institute for Integrative Biology (iBio), Chilean National Agency for Research and Development, Santiago 8331150, Chile.

Abstract:
Melanoma is a highly aggressive skin cancer characterized by dynamic phenotypic switching of malignant melanocytes between proliferative and invasive states. Although this plasticity contributes to tumor progression, metastasis, and drug resistance, the molecular mechanisms governing these transitions remain poorly understood. Accumulating evidence suggests that phenotype switching is associated with the acquisition of stem cell–like properties and is driven by opposing expression patterns of key transcriptional regulators, particularly MITF and BRN2.

Here, we propose a mechanistic mathematical model combined with computational analysis to investigate the dynamical properties of this regulatory system. This approach enables the identification of stable phenotypic states, the transitions between them, and the regulatory factors that promote switch-like behavior. Our framework highlights that melanoma cell fates are not fixed but instead emerge from epigenetic regulatory networks, giving rise to multiple attractor states rather than static outcomes, whose modulation can bias transitions between them. In particular, stabilizing the proliferative phenotype may reduce the emergence of invasive and metastatic states, thereby potentially limiting therapeutic resistance in melanoma.

Poster 26: Plasticity Is Not Just ”Change”: Formalizing the Concept Beyond Semantic Ambiguity

Plasticity Is Not Just ”Change”: Formalizing the Concept Beyond Semantic Ambiguity

Presenter: Yael Daniel Hernandez Gonzalez
Authors: Yael Daniel Hernández-Gonzalez (1,2), Mariana Gómez-Schiavon (2,3)
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3: Millennium Science Initiative Program, Millennium Institute for Integrative Biology (iBio).

Abstract:
Phenotypic plasticity is a central concept in biology, yet it is often used ambiguously to describe a wide range of environmentally induced changes, from stochastic fluctuations to regulated phenotypic transitions. This lack of precision limits our ability to compare different systems and makes it difficult to identify the specific mechanisms that set plasticity apart from other forms of variation. Here, we propose a conceptual framework that defines plasticity as a dynamic and system-level property, rather than a static attribute of phenotypes. We argue that plasticity arises when a biological system reconfigures its internal regulatory dynamics in response to specific cues, allowing access to alternative stable states while preserving organizational integrity. Within this framework, plasticity is distinguished from passive flexibility, homeostasis, and stochastic noise by its regulated nature, state dependence, and capacity for persistence. To illustrate the internal consistency of this definition, we explore a minimal gene regulatory model based on a symmetric toggle switch. In this system, an environmental signal acts by reducing the functional availability of a regulatory protein through rapid sequestration, without altering the network topology or introducing new dynamical variables. This signal deforms the system’s dynamical landscape, inducing transitions between stable expression states. Importantly, transient signals can lead to persistent phenotypic changes via hysteresis, providing a form of memory without explicit genetic or epigenetic encoding. Together, this work emphasizes the importance of formalizing plasticity in dynamical terms and demonstrates that key features of plastic responses can emerge from minimal regulatory architectures. This framework provides a basis for clearer conceptual distinctions and for the development of mechanistic and evolutionary models of plasticity.

Poster 27: Generating phenotypic heterogeneity in fluctuating environments: Between Oscillations and Bistability.

Generating phenotypic heterogeneity in fluctuating environments: Between Oscillations and Bistability.

Presenter: Emmanuel Hernández Sánchez
Authors: Emmanuel Hernández Sánchez (1,2), Mariana Gómez-Schiavon (1,3)
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3: ANID—Millennium Science Initiative Program—Millennium Institute for Integrative Biology (iBio), Santiago, Chile.

Abstract:
Phenotypic transitions and heterogeneity play a central role in the adaptive behavior of biological systems, particularly in fluctuating environments. Oscillatory and bistable dynamical regimes are two fundamental mechanisms capable of generating and sustaining phenotypic heterogeneity; however, determining which of these dynamics governs the behavior of complex biological networks remains a major challenge. Although both regimes can give rise to multimodal phenotypic distributions, they are otherwise fundamentally different: they are mathematically well characterized, arise from distinct minimal network architectures, and exhibit different stochastic dynamics. Their similar observable behavior, combined with intrinsic biochemical noise and limited experimental data, has hindered their reliable identification, particularly in the case of bistability. We propose to address this challenge by systematically comparing oscillatory and bistable regimes in the context of stochastic biological dynamics, focusing on how each mechanism generates phenotypic heterogeneity and responds to fluctuating environments. By analyzing stochastic behavior, multimodality, and dynamical signatures produced by minimal network models under fluctuating conditions, we aim to determine when and why these regimes become difficult to distinguish. Through this framework, our understanding of phenotypic heterogeneity in biological systems will improve, allowing for better interpretation of both experimental and theoretical studies.

Poster 30: Bistability and Transcriptional Bursting: A Comparative Study of Noise-Driven Switching Dynamics

Bistability and Transcriptional Bursting: A Comparative Study of Noise-Driven Switching Dynamics

Presenter: Diego Morales Martínez
Authors: Morales-Martínez, Diego (1) Álvarez-MartÍnez, Roberto (2), Gómez-Schiavon Mariana (3)
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Abstract:
At molecular scales, biological systems are stochastic and this randomness is known as biochemical noise. In the presence of biochemical noise, positive feedback with bistability and transcriptional bursting models exhibit bimodal distributions and switch dynamics; in both cases, these are actually emergent properties of the noise. By analyzing their dynamical properties and noise-driven transitions, we aim to discover how these mechanisms contribute to cellular heterogeneity. In particular, we seek to determine whether there are biologically relevant differences between the two mechanisms—for example, whether a gene regulatory network with positive feedback confers higher fitness than an otherwise identical network driven by transcriptional bursting. Altogether, this comparative framework is intended to assess whether distinct noise-driven regulatory mechanisms can produce similar phenotypic outcomes or instead differentially impact robustness, adaptability, and fitness, despite generating comparable levels of cellular heterogeneity. To evaluate how the dynamics of these two mechanisms differ, we developed three models of genetic circuits: (1) a positive feedback model, which can exhibit bistability, (2) a regulated gene that can display transcriptional bursting, and (3) a hybrid model incorporating both mechanisms. The conditions and properties under which each of these mechanisms displays bimodal distributions will be characterized and compared. As preliminary results, we found that in the deterministic representation the transcriptional bursting model is monostable under all conditions. While the positive feedback and hybrid model depends on the parameter values for determining its global dynamic. In the latter two cases, we observed different behaviors.

Genome Evolution Lab

Responsable: Dra. Lucía Morales

Poster 2: Genomic Insights into Functional Adaptation of Non-Model Yeasts in Traditional Agave Fermentations in Mexico

Genomic Insights into Functional Adaptation of Non-Model Yeasts in Traditional Agave Fermentations in Mexico

Presenter: David Figueroa
Authors: David Figueroa (1), Lucia Morales (1)
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1: Laboratorio Internacional de Investigación sobre el Genoma Humano (LIIGH), Universidad Nacional Autónoma de México, Juriquilla, Mexico.

Abstract:
Traditional agave fermentations in Mexico constitute a complex and dynamic microbial system, shaped by open and non-inoculated processes that promote colonization by locally adapted yeast communities. Recent studies have identified a core group of yeast species recurrently present across distinct agave-producing regions, revealing high genetic diversity and strong geographic structure, particularly in Saccharomyces cerevisiae. However, the genomic diversity and adaptive mechanisms of non-Saccharomyces species associated with this niche remain largely unexplored. To investigate the genetic diversity and functional adaptation mechanisms of yeasts involved in agave fermentations by integrating genomic, phenotypic, and transcriptomic approaches. Genomes from representative isolates of key species (Pichia kudriavzevii, P. manshurica, P. kluyveri, P. teotihuacanensis, and Torulaspora delbrueckii) collected across multiple agave-producing regions in Mexico will be sequenced and annotated. In parallel, we will assess phenotypic performance and transcriptional responses in juices derived from different agave species, revealing the genes and metabolic pathways underlying adaptation to agave juice and distinguishing shared from species-specific adaptive strategies. Overall, this work aims to enhance our understanding of yeast adaptive evolution in traditional fermentations, while generating knowledge with potential biotechnological applications. Keywords: Agave fermentation, non-model yeasts, genomics, functional adaptation

Poster 6: Temporal Dynamics of Yeasts, Bacteria and Viruses During Agave Fermentations

Temporal Dynamics of Yeasts, Bacteria and Viruses During Agave Fermentations

Presenter: Mariana G. Guerrero Osornio
Authors: Mariana Guerrero-Osornio (1), Fernanda Requena-Romo (1), Maritrini Colón (1), Agnès Thierry (2), Eugenio Mancera (3), Alexander DeLuna (4), Martial Marbouty (2), Lucía Morales (1)
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4: Unidad de Genómica Avanzada (Langebio), Centro de Investigación y de Estudios Avanzados del Instituto Politécnico Nacional, Irapuato, Mexico.

Abstract:
Agave fermentations occurring during spirit production are reservoirs of diverse and dynamic microbial communities. These traditional processes take place in open tanks, where yeasts are know to be key players in the fermentation. In this study, we tracked the progression of a single agave fermentation using the high-resolution Meta-HiC technique. This method combines

metagenomics with Chromosome Conformation Capture to recover high-quality Metagenome-Assembled Genomes (MAGs) from the most abundant species across multiple domains of life, unveiling the genomes of bacteria, viruses, and their hosts, that coexist with yeasts throughout the fermentation process. Among the identified microorganisms, the bacterium Zymomonas mobilis was the one with the highest relative abundance that kept increasing over the course of fermentation. The high quality of its MAG revealed genes not found in other Z. mobilis genomes, potentially contributing to its adaptation to this environment. Based on the microbiome data, the predominant fungal species was the yeast Saccharomyces paradoxus, which dominated the fungal community throughout the fermentation process. Viral MAGs were consistently detected, and host prediction analyses revealed dynamic phage–bacteria interactions, suggesting that viruses also play a ecological role during fermentation. By applying Meta-HiC across the fermentation, we achieved unprecedented resolution in genome reconstruction of microbial players, enabling the discovery of the genetic components that drive adaptation to this complex environment.

Poster 11: A MetaHiC-Based Pipeline for Detecting Yeast Hybrids
A MetaHiC-Based Pipeline for Detecting Yeast Hybrids
Presenter: Maria Fernanda Requena Romo
Authors: Fernanda Requena-Romo(1,2), Mariana Guerrero-Osornio(1,3), Lucia Morales(1)
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3: Posgrado en Ciencias Biológicas, Universidad Nacional Autónoma de México.

Abstract:
Previous studies have shown that hybridization between Saccharomyces cerevisiae and Saccharomyces paradoxus is common in the fermentation environments of agave spirit distilleries. However, the timing of these hybridization events and their impact throughout the fermentation process remain unclear. To address this, we developed a MetaHiC-based pipeline to detect and characterize hybrids at different stages of fermentation. To track hybrids during fermentation, we developed a MetaHiC-based pipeline to detect physical interactions between S. cerevisiae and S. paradoxus genomes. MetaHiC libraries were prepared every 12 hours from the initial inoculum to the end of fermentation, yielding six timepoints for analysis. Although no hybrids were detected, the pipeline consistently identified a strong interaction between an 16 Kb region of S. cerevisiae and the S. paradoxus genome, indicating the sustained presence of an S. paradoxus strain harboring an introgression in chromosome 8. This result provides a proof of concept for the pipeline's capacity to detect genomic introgressions and interspecific hybrids from MetaHiC data, highlighting its potential for broader applications in future studies involving suspected hybridization events.

Poster 17: Bidirectional gene flow in populations of S. paradoxus and S. cerevisiae from agave fermentations
Bidirectional gene flow in populations of S. paradoxus and S. cerevisiae from agave fermentations
Presenter: Iván Sedeño
Authors: Iván Sedeño (1,2), J Abraham-Avelar Rivas(3), Eugenio Mancera (4), Alexander DeLuna (3), Lucía Morales(1)
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4: Departamento de Ingenieria genética (CINVESTAV).

Abstract:
Gene flow is a key evolutionary force in the Saccharomyces genus, where hybridization has prevailed, leaving signatures of introgression in natural populations. In recent years, such events

have been increasingly documented in both wild and domesticated yeast populations, with important implications for their adaptation, ecological range and evolutionary history. In this study, we build on our previous work by comparing earlier findings of Saccharomyces paradoxus introgressions into S. cerevisiae strains from agave fermentations with new evidence showing that gene flow has also occurred in the opposite direction, albeit resulting in fewer introgressed genes in agave-associated S. paradoxus isolates. Our current work aims to determine the prevalence of these introgressions across clades and to investigate their adaptive roles and functional significance. These results underscore the permeability of species boundaries within the Saccharomyces genus and open new avenues for exploring the evolutionary and ecological consequences of interspecific gene flow in both wild and domesticated yeast populations.

Poster 29: Genomic and Transcriptomic Characterization of a Saccharomyces cerevisiae × Saccharomyces paradoxus Hybrid Yeast Strain
Genomic and Transcriptomic Characterization of a Saccharomyces cerevisiae × Saccharomyces paradoxus Hybrid Yeast Strain
Presenter: Diego Alejandro Marquez Cerna
Authors: Diego Alejandro Marquez Cerna (1,2), Samuel Quiñones Galeana (1)
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2: Arkansas State University Department of Biological Sciences.

Abstract:
Interspecific Saccharomyces Hybrids integrate divergent parental genomes, providing a powerful system to study genome architecture and gene expression regulation under different environmental growth conditions. However, the complexity of hybrid genomes limits precise characterization of subgenome contributions. In this ongoing project, long-read sequencing and transcriptomic data are being integrated to generate a de novo annotated assembly of a hybrid S. cerevisiae x S. paradoxus Using Oxford Nanopore reads, a hybrid genome assembly is being constructed to identify regions of loss of heterozygosity and define orthologous gene pairs between both subgenomes based on this framework, gene expression is being evaluated in cultures grown in minimal medium, Agave salmiana juice, and sorghum hydrolysate Analyses include comparisons of 1:1 orthologous genes and allele-specific expression to explore patterns of parental expression dominance overall, this work aims to establish a genomic and trancriptomic framework to undersyad how different usbtrates modulate the functional contribution of each subgenome in biotechnological relevant hybrid

Mendelian Genomics and Precision Health Lab

Responsable: Dra. Claudia Gonzaga Jauregui

Poster 5: Genomic findings and SHANK 3 microduplication in a patient with autism spectrum disorder
Genomic findings and SHANK 3 microduplication in a patient with autism spectrum disorder
Presenter: Valeria Gómez-Vela
Authors: Valeria Gómez-Vela(1,2), Renée Barreda-Fierro(3), Claudia Gonzaga-Jauregui(1,3)
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Abstract:
Autism spectrum disorder (ASD) is a group of neurodevelopmental conditions, characterized by deficits in social communication and interactions, as well as repetitive and restrictive behaviours, with or without other features like intellectual disability or seizures. ASD affects 1 percent of the global population and in Mexico 45,000 children are diagnosed with the condition. ASD is clinically and genetically heterogeneous with more than 100 genes involved in diverse neuronal biological pathways being associated with the disorder. Genomic technologies, such as exome sequencing (ES) and chromosome microarray analysis (CMA), have proven to identify the molecular causes of genetically heterogeneous disorders such as autism. The SRC homology 3 (SH3) and multiple Ankyrin repeat domain (SHANK) family of proteins are known as organisers of excitatory glutamatergic synapses. SHANK3 is one of the three known members of the family and it is abundantly expressed in the cortex, hippocampus, and olfactory bulb. Phelan Mc-Dermid syndrome (PMS) is a rare neurodevelopmental disorder caused by loss of SHANK3 through deletion of the 22q13 region or by pathogenic loss-of-function variants in the gene. PMS is characterized by global developmental, motor, and speech delay with moderate to severe intellectual disability and autistic behaviors, among other features. Conversely, duplications of the 22q13 region encompassing SHANK3 have been reported in the literature in a few patients with ASD, mild developmental delay, behavioral problems and attention-deficit/ hyperactivity disorder . Here, we report a 14 year old male patient referred for clinical features of ASD, developmental language disorder, hyperkinetic disorder, hypoalgesia and sleep 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. The clinical presentation of our patient is consistent with the features reported in 22q13 duplication syndrome patients providing a molecular diagnosis for the patient and adding to the characterization of this rare genomic disorder.

Poster 20: Identification of disease-associated variants in Heme production genes associated with Porphyrias

Identification of disease-associated variants in Heme production genes associated with Porphyrias

Presenter: Román Cervantes Levario
Authors: Román Cervantes Levario (1)(2), Valeria Gómez Vela (1)(2), Claudia Gonzaga Jauregui (1)
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2: Genomic Sciences Undergraduate Program, Licenciatura en Ciencias Genómicas, ENES Juriquilla, Universidad Nacional Autónoma de México, UNAM Campus Juriquilla, Querétaro, México.

Abstract:
Porphyrias are a group of rare genetic diseases in which different intermediate metabolic products in the biosynthetic pathway of the heme group of hemoglobin, known as porphyrins, accumulate and become toxic to the nervous or other systems. Porphyrias can be classified into three groups based on their clinical presentation: 1) Acute Hepatic Porphyrias (AHPs) characterized by neurovisceral attacks, insomnia, fatigue, abdominal pain, tachycardia, hypertension, and seizures; 2) Cutaneous Hepatic Porphyrias (CHPs), where patients may develop blisters on sun-exposed skin with liver damage and iron overload; and Erythropoietic Cutaneous Porphyrias (ECPs) where patients present with photosensitivity, blistering, and anemia. In AHPs, attacks can be induced by environmental triggers such as stress, alcohol, or certain drugs; whereas for cutaneous porphyrias sun exposure should be avoided. While few therapeutic options are available for porphyria patients, avoiding triggers and exposures for individuals at risk is the main management approach. There are nine genes involved in heme biosynthesis, eight of which, when

altered, result in porphyrias. Pathogenic variants in the genes ALAD, HMBS, UROS, UROD, CPOX, PPOX, and FECH that result in reduced or loss-of-function of the encoded enzymes lead to the accumulation of porphyrins. Conversely, gain-of-function variants in the gene ALAS2 result in increased enzyme activity and porphyrin accumulation. To date, more than 1,000 variants causing the various types of porphyrias have been reported. In this study, we aim to identify pathogenic and likely pathogenic variants in porphyria associated genes in the Mexican population, leveraging genomic data available from the Mexico City Prospective Study (MCPS). We will characterize the variant and allele frequency spectrum associated with these diseases and estimate the carrier frequency and genetic prevalence of porphyrias in this population. This information may help develop public health strategies for carrier screening, early detection, and proper management of porphyrias in Mexico.

Poster 25: Modelado funcional en Caenorhabditis elegans de variantes genéticas en el complejo PP2A asociadas al Síndrome de Houge-Janssens
Modelado funcional en Caenorhabditis elegans de variantes genéticas en el complejo PP2A asociadas al Síndrome de Houge-Janssens
Presenter: Kitzia Gómez Cepeda
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Abstract:
La fosforilación de proteínas ocurre principalmente en los aminoácidos serina (Ser) y treonina (Thr), representando aproximadamente el 96% de los eventos de fosforilación y constituyendo la modificación post-traduccional más común. Los complejos de proteínas fosfatasas 1 (PP1) y 2A (PP2A) son las principales fosfatasas de Ser/Thr en la célula, siendo PP2A la de mayor actividad. El complejo PP2A regula la señalización y fisiología celular, y es esencial durante el desarrollo y funcionamiento del cerebro. PP2A está compuesto por una subunidad estructural (PPP2R1A), una regulatoria (PPP2R5D) y una catalítica (PPP2CA). Mutaciones de cambio de aminoácido, generalmente de novo, en los genes que codifican estas subunidades han sido asociadas con el Síndrome de Houge-Janssens (SHJ), un desorden del neurodesarrollo humano caracterizado por retraso en el desarrollo global, discapacidad intelectual variable e hipotonía, entre otras manifestaciones clínicas. La secuenciación genómica de pacientes con desórdenes del neurodesarrollo ha permitido identificar variantes raras probablemente patogénicas que en muchos casos permanecen sin significancia clínica certera debido a la falta de información funcional sobre su efecto. Mediante técnicas de ingeniería genética y biología molecular, en este estudio se generarán en el modelo Caenorhabditis elegans mutaciones puntuales equivalentes a variantes identificadas en pacientes con SHJ, introducidas en los genes ortólogos del nemátodo. Se evaluarán los fenotipos de animales mutantes, tales como cambios en tamaño, morfología, función motora, comportamiento u otros rasgos, en comparación con controles no modificados, a fin de determinar el efecto de las variantes. Finalmente, se analizará la actividad neuronal específica mediante técnicas de imagenología en animales vivos, utilizando proteínas fluorescentes como reporteros. Esta estrategia permitirá determinar la funcionalidad de variantes de significado incierto en genes asociados a desórdenes del neurodesarrollo en un modelo in vivo, contribuyendo a una mejor comprensión de los mecanismos moleculares y celulares que subyacen a estas enfermedades neurológicas.

Poster 33: In vivo functional modeling of SMARCA5 variants associated with a novel neurodevelopmental disorder in Caenorhabditis elegans
In vivo functional modeling of SMARCA5 variants associated with a novel neurodevelopmental disorder in Caenorhabditis elegans
Presenter: Ariana Gabriela Reyes Silva

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Abstract:
Neurodevelopmental disorders are a group of conditions characterized by cognitive, behavioral, and developmental impairments due to altered processes in the early development of the central nervous system. These disorders are often associated with pathogenic genetic variants that disrupt critical biological pathways. Recently, the chromatin-remodeling gene SMARCA5, which encodes the SNF2H protein and plays a key role in the regulation of gene expression during development, has been associated with a novel neurodevelopmental disorder, although its precise role in neurodevelopment remains incompletely understood. Pathogenic missense variants in SMARCA5 were identified in 12 probands from 10 families from around the world sharing a phenotype characterized by microcephaly, short stature, and mild developmental delay. Additionally, a de novo missense variant was identified in a Mexican patient presenting a similar phenotype. However, to better understand the effect of newly identified variants in patients and the biological function of the gene, experimental functional studies in vitro or in vivo are necessary. This project aims to functionally model likely pathogenic missense variants identified in patients with SMARCA5-associated neurodevelopmental disorder in the model organism Caenorhabditis elegans. Using CRISPR-Cas9 genome editing, human SMARCA5 variants are being introduced into the worm ortholog, isw-1, which shares a 63% identity and 77% similarity with the human gene. Phenotypic analyses of genetically modified animals will aim to assess the functional consequences of these variants in vivo. Our experiments and analyses will provide insights into the molecular and cellular mechanisms underlying SMARCA5 function and its role in neurodevelopment. By integrating genetic editing and phenotypic analyses, this project seeks to bridge the gap between genetic discovery and functional understanding of missense variants associated with SMARCA5-associated neurodevelopmental disorder.

Poster 35: Panorama actual del tamizaje neonatal genómico para la detección temprana de enfermedades genéticas en el mundo
Panorama actual del tamizaje neonatal genómico para la detección temprana de enfermedades genéticas en el mundo
Presenter: Erandi Abril Salas Romero
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Abstract:
La implementación de la secuenciación genómica para el diagnóstico certero y temprano de enfermedades de origen genético ha mejorado significativamente el pronóstico y calidad de vida de pacientes con estas enfermedades. En los últimos años, estudios de investigación se han iniciado alrededor del mundo para evaluar la aplicabilidad y efectividad de la secuenciación genómica para tamizaje neonatal a escala poblacional. Se realizó una búsqueda exhaustiva de la literatura sobre proyectos de tamizaje neonatal genómico (TNG) a nivel mundial con el fin de evaluar los criterios utilizados para la selección de genes candidatos e identificar los hallazgos más relevantes. Se llevó a cabo una revisión manual de las listas de genes disponibles reportadas en cada estudio, se recopiló información relevante sobre dichos genes y las condiciones genéticas a las que se les ha asociado, así como información sobre tratamientos disponibles.

Se recopiló una lista de 1,824 genes provenientes de 26 proyectos de TNG distintos que se llevan a cabo en 15 países. Se observó que 70% de las condiciones asociadas a estos genes inicia en la infancia; 66% tienen tratamiento disponible en México. Del total de genes, 53% (n = 961) tienen una relación gen-enfermedad “definitiva” y 49% (n = 904) presenta una penetrancia “alta” de las condiciones asociadas. Entre los principales hallazgos de los estudios que han reportado resultados destaca una tasa de diagnóstico del 1.42%, reducción de la morbimortalidad, así como costo-efectividad respecto a vías diagnósticas tradicionales. La evidencia disponible sobre la implementación de TNG en diferentes países ha demostrado mejores desenlaces en salud, prometiendo ampliar sustancialmente la detección temprana de enfermedades genéticas accionables. Sin embargo, el alto costo y acceso limitado a tecnologías de secuenciación genómica en países de recursos medios y bajos ha limitado su aplicación en países como México y otros países de Latinoamérica.

Paleogenomics and Evolutionary Biology Lab

Responsable: Dr. Federico Sánchez Quinto

Poster 16: Analysis of a deeply-phenotyped familial hypercholesterolemia cohort from Mexico shows a role for both rare and common alleles across known dyslipidemia genes and reveals structural variation in a novel locus

Analysis of a deeply-phenotyped familial hypercholesterolemia cohort from Mexico shows a role for both rare and common alleles across known dyslipidemia genes and reveals structural variation in a novel locus

Presenter: Edgar Pavel Salazar Fernández

Authors: Nicholas Katsanis (1,2), Niki Mourtzi (1), Consuelo D Quinto-Cortés (1), Alexandro J Martagon (6,7,8), Alexander G Ioannidis (1,3,9,10), Francisco M De La Vega (1,3), Jeff Gulcher (1), Ming Ta Michael Lee (1), Mohammad A Faghihi (1), Arturo Lopez-Pineda (1,4,5), Sonia Moreno-Grau (1), Daniel Mas Montserrat (1,3), Míriam Barrabés (1,3), David Bonet (1,3), Pavel Salazar Fernandez (1,4), Jeff Wall (1), Babak Moatamed (1), Roopa Mehta (6,11), Gabriela A Galan-Ramirez (6), Rafael Zubirán (6), Daniel Elias-Lopez (6,11), Teresa Tusié-Luna (11), Carlos A Aguilar-Salinas (6,7,11,12), Carlos D Bustamante (1,3,10); F. H. Mexican Registry Group

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Abstract:

Familial hypercholesterolemia (FH) is a genetic disorder driven in part by mutations in three

genes that encode components of the cholesterol pathway: LDLR, APOB, and PCSK9. However, the majority of FH genetics has been performed in individuals of European descent. Here, we leveraged a cohort of 300 patients from the Mexican FH registry to understand how rare, high liability alleles and common variants might contribute to shaping individual risk. Using a combination of whole exome and of short- and long-read whole genome sequencing, we report three key findings. First, we observed that rare pathogenic point mutations and structural variants in all known FH genes, together with variants in APOE, CREB3L3, and PLIN1, contribute to a molecular FH diagnosis in 67% of families, including novel gene-disruptive copy number variants (CNVs) which arose in a native American background. Second, ancestry-adjusted polygenic risk score analysis identified a significant liability for coronary artery disease, hypertension, LDL, HDL, and Type 2 Diabetes. The polygenic signal for LDL was present in patients with rare, pathogenic FH mutations and was more prominent in individuals bereft of a molecular FH diagnosis. Finally, we report both a whole-gene duplication and common, non-coding variants in a novel locus, PDZK1, which contribute to the genetic burden of FH, a finding we replicated in the UK Biobank (UKB). Together, our analyses illustrate the value of genetic studies in non-European populations and reinforce the notion that individual risk to disease can arise from both rare, large effect alleles (alone or in combination across genes) and common variants that increase the mutational burden of a biological system.

Poster 21: Análisis de las afinidades genéticas y estructura social de los individuos prehispánicos excavados en Tlatel 7 de Santa Lucía I
Análisis de las afinidades genéticas y estructura social de los individuos prehispánicos excavados en Tlatel 7 de Santa Lucía I
Presenter: Leonardo Yair Correa Mendoza
Authors: Leonardo Yair Correa Mendoza (1), Alejandra Castillo Carbajal (1), Ruben Manzanilla López (2), María del Carmen Ávila Arcos (1),Juan Carlos Equihua Manrique (2), Federico Andrés Sánchez Quinto (1)
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2: Dirección de Salvamento Arqueológico, Ciudad de México, México, Instituto Nacional de Antropología e Historia, México.

Abstract:
Durante el Epiclásico (600–900 d.C.), tras la caída de Teotihuacan, el noreste de la Cuenca de México se caracterizó por asentamientos aldeanos dispersos en torno a pequeños centros políticos. Este contexto arqueológico sugiere una intensa movilidad poblacional, así como inestabilidad política y conflicto. En este proyecto se extrajo, secuenció y analizó ADN antiguo de seis individuos de Tlatel 7 del sitio Santa Lucía (Estado de México), asociados con el periodo Epiclásico, con el objetivo de caracterizar su historia demográfica y la estructura social de dichos individuos. Este estudio generó los genomas mitocondriales de los seis individuos analizados, y datos nucleares de tres de ellos. Se caracterizaron los linajes mitocondriales y se evaluaron vínculos de parentesco para las tres muestras con datos nucleares. Asimismo, se analizaron las afinidades y estructura genética de los individuos del Tlatel 7, en comparación con datos genéticos de otros individuos prehispánicos de referencia del Centro de México y muestras indígenas contemporáneas para investigar su historia demográfica. Los resultados sugieren que los tres individuos con datos autosómicos no estaban emparentados entre sí, y que los individuos de este entierro tenían perfiles demográficos similares a muestras antiguas y contemporáneas del Centro de México. Los siguientes pasos serán buscar recuperar una mayor cantidad de ADN de estás muestras de este sitio arqueológico y otros yacimientos cercanos, para identificar si desplazamientos poblacionales importantes, como flujo génico o cuellos de botella poblacionales, estuvieron asociados con el periodo de inestabilidad social inferido a partir del registro arqueológico en esta zona del México prehispánico.

Poster 31: Molecular sexing of ancient faunal remains: methods, limits and prospects
Molecular sexing of ancient faunal remains: methods, limits and prospects

Presenter: Rigoberto Padilla Bustos
Authors: Rigoberto Padilla-Bustos (1), Federico Sánchez-Quinto (1)
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Abstract:
High-resolution identification of biological sex from archaeological and palaeontological remains is central to reconstructing social structure, demography, and behaviour in past human and animal populations, yet traditional osteological sex-determination is often unreliable for fragmentary or weakly dimorphic specimens. The advent of ancient DNA and whole-genome sequencing (WGS) has provided a powerful alternative to accurately determine chromosomal sex from genomic data. While several WGS-based strategies have been implemented to infer sex karyotypes in XY systems from ancient DNA data (i.e. Ry, X-to-autosome, Rx and karyo_RxRy methods), there is not a clear consensus on the minimal sequencing data threshold needed to obtain reliable results using most of them. Here we evaluated the performance of these methods by subsampling >1x genomes across 15 samples from 5 vertebrate families to files harbouring different sequencing reads counts (20K,10K, 5k, 2K, etc), representing ultra-low coverage levels, and assessed their sex karyotypes at each coverage level. Our results revealed the minimal sequencing reads count threshold needed to obtain a >95% concordance sex determination estimate result for each analyzed method. Our study sheds light on the limits of sex determination using ancient DNA, which is of great relevance for researchers working with poorly preserved samples, often deriving from subtropical and tropical latitudes.

Poster 32: Identificación taxonómica de murciélago desconocido utilizando datos genómicos.
Identificación taxonómica de murciélago desconocido utilizando datos genómicos.
Presenter: Natalie Berenice Pineda Morán
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Abstract:
El género Eumops comprende murciélagos microquirópteros (familia Molossidae), también conocidos como murciélagos de bonete o mastín. Este género se distribuye desde el sur de Estados Unidos de América hasta Sudamérica. Dentro de este género, el complejo Eumops glaucinus incluye a las especies: E. floridanus, E.ferox, E. underwoodi, entre otras, las cuales se distribuyen principalmente desde el sur de México hacia el resto del continente, incluyendo algunas islas del Caribe. En 2022 en Saltillo (Coahuila), se descubrió un espécimen de murciélago previamente desconocido. Su morfología craneal y postcraneal indican pertenencia al género Eumops. Sin embargo, la muestra presenta variación morfológica atípica, incluyendo algunos caracteres ancestrales. Adicionalmente, la presencia de Eumops no ha sido descrita aún en el noreste de México. Estas observaciones sugieren que quizá este espécimen podría pertenecer a una especie de Eumops. El presente estudio tiene como objetivo obtener el genoma mitocondrial de la muestra problema para investigar su afinidad filogenética con otras especies de murciélagos, y evaluar la hipótesis del descubrimiento de una nueva especie. La metodología desarrollada al momento, involucra tres partes. Primero se extrajo DNA de un pedazo de costilla, se construyeron librerías de secuenciación y se generaron datos de secuenciación exploratorios. Paralelamente, se ensambló el genoma mitocondrial de una muestra E.

floridanus de EUA para generar un genoma mitocondrial de referencia al cual mapear los datos. Así mismo, se descargaron de NCBI tres mitogenomas de las especies Tadarida brasiliensis, Myotis myotis y Molossus molossus como referencia. Un mapeo competitivo de la muestra problema contra las diferentes referencias mitocondriales sugiere una mayor cercanía genética con T.brasiliensis que con E. floridanus. Los siguientes pasos del proyecto involucran revisar el ensamble mitocondrial generado, y obtener más mitogenomas referencia de Eumops a lo largo de su distribución geográfica en el continente para evaluar la hipótesis planteada.

Population and Evolutionary Genomics Lab

Responsable: Dra. María C. Ávila Arcos

Poster 1: Tracing Introduced Pathogen Lineages and Social Dynamics in Colonial Mexico City through Paleogenomics

Tracing Introduced Pathogen Lineages and Social Dynamics in Colonial Mexico City through Paleogenomics

Presenter: Laura Carrillo Olivas

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Abstract:

European colonization of the Americas led to profound demographic shifts and epidemic outbreaks, yet the molecular diversity of pathogens affecting marginalized colonial populations in Mexico remains poorly understood. We present a large-scale paleogenomic analysis of colonial infections in Mexico, examining 95 individuals and seven dental calculus samples from the Hospital Real San José de los Naturales and Templo de la Inmaculada Concepción in Mexico City.

Mitochondrial DNA and autosomal ancestry analyses revealed predominantly Indigenous genetic ancestry, alongside individuals of African origin, as well as others with Indigenous-African and Indigenous-European admixture, highlighting the multiethnic nature of these marginalized populations. Molecular sex analysis revealed balanced XX and XY ratios, with one individual showing a rare XYY karyotype. To identify ancient pathogens, we developed a novel scoring system combining taxonomic classification and the presence of virulence factors, which revealed a suite of introduced microbes. We report the first ancient genomic identification of Streptococcus pyogenes, alongside an Afro-Asian lineage of Streptococcus pneumoniae. Moreover, we find evidence of co-circulation of both non-typhoidal and typhoidal fever-associated Salmonella enterica lineages, a combination yet undocumented in the Colonial Americas, alongside an infection by Citrobacter freundii and oral pathogens such as Tannerella forsythia, Porphyromonas gingivalis, and Treponema denticola. Our analyses uncover an interesting divergence in pathogen distribution. Oral pathogens were ubiquitous across all genetic ancestries, indicating multiethnic widespread community transmission, while potentially severe systemic infections were mainly concentrated among non-admixed Indigenous individuals. These patterns suggest how colonial social structures and violence could have created differential health risks, with Indigenous populations being the most vulnerable. Our findings provide a historical framework for understanding the initial introductions and ecological drivers that fueled health disparities, offering insight into the long-term evolution of pathogens in societies impacted by colonialism.

Poster 3: Ancient virus identification through ancient DNA in prehispanic and colonial human remains in Mexico

Ancient virus identification through ancient DNA in prehispanic and colonial human remains in Mexico

Presenter: Miguel Ángel Flores Varela
Authors: Miguel Ángel Flores-Varela(1), Anahí Tania Sánchez-León(1), Laura Carrillo-Olivas(1), Alejandra Castillo-Carbajal(1), Elizabeth Mejía Pérez Campos(2), María C. Ávila-Arcos(1), Daniel Blanco-Melo(3)
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Abstract:
The Spanish conquest of Mexico in the XVI century led to a bidirectional exchange of microorganisms between Indigenous American populations and Europeans. The early colonial period was therefore marked by severe epidemics that caused a dramatic demographic collapse. The Hospital San José de los Naturales (HSJN), in Mexico City, was a key institution that attended infected Indigenous individuals during this time. Previous work from our group recovered three ancient genomes of parvovirus B19 and one of hepatitis B virus from this and another Colonial site from Mexico City, namely “La Conchita” , a Colonial chapel. To further explore the presence of these and other viruses, we analyzed additional samples, 21 from La Conchita, 82 from HSJN and 11 from the pre-Hispanic site of Toluquilla. The inclusion of Toluquilla aimed to assess whether viruses associated with colonial epidemics were present prior to the arrival of Spaniards and to provide an initial overview of viral diversity in prehispanic populations. Standard ancient DNA (aDNA) pipelines were applied, including metagenomic screening using MALT and CZID. Subsequently, targeted capture-enrichment assays were performed to increase ancient viral DNA abundance, followed by mapping against viral reference genomes. Our analyses yielded positive results for parvovirus B19 in several HSJN samples, as well as preliminary signals compatible with variola virus that warrant further capture-enrichment analyses. In contrast, prehispanic samples showed a low abundance of pathogenic viral DNA, which may reflect limitations in viral DNA preservation, incomplete reference databases, or a genuinely lower prevalence of pathogenic viruses prior to the colonial period.

Poster 4: Genomic history and ancestral origins of Afro-Mexican populations
Genomic history and ancestral origins of Afro-Mexican populations
Presenter: María Fernanda García Rodríguez
Authors: María Fernanda García Rodríguez (1, 2), Shreya Ramachandran (3), María José Palma (4), Juan Esteban Rodríguez-Rodríguez (5), Isabella S. Gandara (6,7) , Marcus Feldman (8), Carlos D. Bustamante (9), Andres Moreno-Estrada (10), Cesar A. Fortes-Lima (11), Marcela Sandoval (4), Julie Wesp (12), María C. Ávila-Arcos (1).
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Abstract:
The Mexican population has three principal ancestries: Indigenous American, European, and African. Although the African ancestral component is proportionally smaller at the population level, it holds significant importance in understanding the country's genetic diversity. However, few studies have explored the genetic contributions of Afro-descendant communities in Mexico. Our research aims to characterize and analyze the genetic structure of Afro-Mexican populations, tracing their genetic origins at the subcontinental level, both within Africa and Mexico. We conduct genomic analyses of volunteer participants from Veracruz and the Costa Chica regions of Guerrero and Oaxaca, alongside implementing methods for detailed local ancestry analyses. Preliminary results suggest that the African component in Afro-Mexicans can be traced to two different sources within Africa, while the Indigenous American component clusters with reference populations near their geographical location. The main goal of this research is to highlight the fundamental contribution of Mexico's Afro-descendant populations to our cultural and genetic diversity.

Regulatory Genomics and Bioinformatics Lab

Responsable: Dra. Alejandra Medina Rivera

Poster 15: Factores clínicos asociados a nefritis lúpica en población mexicana.
Factores clínicos asociados a nefritis lúpica en población mexicana.
Presenter: Cielo del Carmen Rodríguez Molina
Authors: Rodríguez-Molina Cielo del Carmen (2), Bravo-García María Fernanda (1), Vera del Valle Sandra Valentina (3), Hernández-Ledesma Ana Laura (1), Peña-Ayala Angélica

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Abstract:
Introducción La nefritis lúpica (NL) representa una de las complicaciones más graves del lupus eritematoso sistémico (LES), asociada con mayor morbilidad, mortalidad y riesgo de daño renal progresivo. En población mexicana, la evidencia sobre factores clínicos e inmunológicos asociados a su presencia es limitada, lo que dificulta la detección temprana y el manejo oportuno. Objetivo Identificar variables clínicas e inmunológicas asociadas a la presencia de nefritis lúpica en población mexicana con LES Metodología Se realizó un estudio transversal y exploratorio utilizando datos del Registro Mexicano de Lupus (LupusRGMX). La nefritis lúpica se definió con base en el diagnóstico clínico registrado. Se analizaron variables clínicas e inmunológicas, incluyendo proteinuria, niveles bajos de complemento (C3 y C4) , edad al diagnóstico, edad actual, uso de corticosteroides y antimaláricos, así como la actividad de la enfermedad, evaluada mediante el índice SLEDAI. El análisis estadístico se efectuó en el software de R (versión 4.5.0), utilizando un modelo de regresión binomial, estimando odds ratios (OR) e intervalos de confianza al 95% . Resultados De los 268 pacientes incluidos, el 96.6% fueron mujeres, con una edad media de 37.8 ± 10.7 años. La prevalencia de nefritis lúpica fue del 19.8%. La presencia de proteinuria (OR:1.92; IC95%:1.59-2.35; p<0.001) y los niveles bajos de complemento C3 y C4 (OR:2.25; IC95%:1.05-4.92; p=0.039) se asociaron significativamente con la presencia de NL. Conclusión En esta cohorte mexicana la presencia de proteinuria y niveles bajos de complemento se identificaron como factores clínicos que contribuyen de manera significativa al desarrollo de nefritis en personas con lupus. Su monitoreo sistemático podría mejorar el diagnóstico, pronóstico y manejo clínico, contribuyendo a prevenir daño renal irreversible y a optimizar la calidad de vida de los pacientes con LES.

Poster 23: Analysis of Genetic Variants Associated with Parkinson's Disease and Depression

Analysis of Genetic Variants Associated with Parkinson's Disease and Depression

Presenter: Diana Delgado Gutiérrez
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Abstract:
BACKGROUND: Parkinson's disease (PD) is a neurodegenerative disease characterized by motor and non-motor symptoms. Depression is a fairly common nonmotor symptom of PD and it impacts the quality of life of patients as well as their caregivers. Studies have shown association of polymorphisms in the brain-derived neurotrophic factor (BDNF) gene with an increased risk of developing depression in individuals with PD. OBJECTIVE: The objective of this study is to analyse the influence of polymorphisms in the BDNF gene on the presence of depression in a European cohort of individuals with PD. METHODS: Genomic data was used from the Global Parkinson's Genetic Program (GP2) along with the Beck Depression Inventory (BDI) score. The sample included 695 individuals with PD, 137 were classified as depressed cases (BDI score >=14) and 558 non-depressed controls. Single-gene analyses of BDNF were performed for all variants and coding variants using a generalized linear model adjusted for age, sex and the first five principal components as covariates, as well as an unadjusted model with PLINK2.0. Burden tests using Sequence Kernel Association Test and Optimal Sequence Association Test

were carried out using previous covariates and coding variants of BDNF with RVTest. RESULTS: Single gene analyses identified 36 significant (p-value < 0.05) variants using all variants and 5 significant variants using only coding variants in BDNF. Upon adjusting for covariates, 5 coding variants were identified in BDNF. CONCLUSIONS: The results suggest the association of coding and non-coding variants in the BDNF gene with depression in individuals with PD, contributing to our understanding of the genetic factors of depression in PD and possibly guide treatment research. This analysis will be replicated in another European cohort using the Geriatric Depression Scale to assess depression, as well as in a Latin American cohort . KEYWORDS: Parkinson's disease, depression, genetic association

Poster 24: Cognitive Impairment in Parkinson's Disease: Association between White Matter Lesion Volume and MoCA Score

Cognitive Impairment in Parkinson's Disease: Association between White Matter Lesion Volume and MoCA Score

Presenter: Mariana Fraustro Méndez

Authors: Mariana Fraustro-Méndez (5), Juan Manuel Esquivias-Farías (1), Yamil Matuk-Pérez (2), Paula Reyes (3), Alejandra Lázaro-Figueroa (4), Carlos Ponce-Fernández (5), Erick Humberto Pasaye (1), César Arturo Domínguez-Frausto (1), Miguel E. Rentería (6), Alejandra Medina-Rivera (3), Alejandra Evelyn Ruiz-Contreras (4), Sarael Alcauter (1).

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Abstract:

Introduction: Parkinson's disease (PD) is a neurodegenerative disorder characterized by progressive loss of dopaminergic neurons in the basal ganglia, specifically in the substantia nigra. It is the most common neurodegenerative disease after Alzheimer's Disease. Bradykinesia, rigidity, and rest tremor constitute the cardinal motor features observed in PD patients. Concurrently, memory impairment represents one of the primary non-motor dysfunctions associated with the condition. Objective: To investigate the association between white matter lesion and global cognitive function in a cohort of Mexican patients with PD. Methods: A cohort study was conducted on 71 Mexican patients with PD (42 males; mean age 65.2 ± 10.8 years). Cognitive function was assessed using the Montreal Cognitive Assessment (MoCA). Structural brain images were acquired on a 3 T Magnetic Resonance Imaging scanner (MRI), including T1 and T2-FLAIR weighted images. The T1w and FLAIR images were processed using the DeepLesionBrain tool, an automated deep-learning pipeline to identify and quantify the volume of white matter (WM) lesions. The Lesion Burden was quantified as the proportion of lesion volume relative to the volume of normal□appearing white matter. Finally, WM lesions volumes were entered as predictors of MoCA score in a General Linear Model adjusted for age, sex, and years of education. Results: Periventricular lesions were the most frequent WM lesion subtype, observed in 78.9% of participants, compared to 15.5% with juxtacortical and 4.2% with deep white matter lesions. In the adjusted model, total WM lesion burden was a statistically significant negative predictor of MoCA ($\beta = -1.867$, $p = 0.042$). When decomposed by region, the periventricular lesion burden (PLB) specifically drove the association ($\beta = -2.178$, $p = 0.024$), indicating that a larger periventricular lesion volume is strongly associated with lower global cognitive scores.

Conclusion: The findings from this cohort demonstrate a negative association between the volume of lesions in the periventricular white matter and global cognitive function of patients with PD. These findings suggest that periventricular WM lesions may serve as an imaging biomarker of cognitive decline in PD.

Poster 37: INFLUENCIA DE LOS FACTORES REPRODUCTIVOS EN LA EDAD DE INICIO DE LA ENFERMEDAD DE PÁRKINSON EN MUJERES MEXICANAS.
INFLUENCIA DE LOS FACTORES REPRODUCTIVOS EN LA EDAD DE INICIO DE LA ENFERMEDAD DE PÁRKINSON EN MUJERES MEXICANAS.

Presenter: Karen Astrid Zárate Saldierna
Authors: Karen A. Zárate-Saldierna (1), Alexis Steven-Garcia (1), Alejandra Lázaro-Figueroa (2), Ulises Caballero-Sánchez (2), Juan Manuel Esquivias-Farías (3), Eugenia Morelos-Figaredo (4), Carlos M. Guerra-Galicia (5), Ingrid Estrada-Bellmann (6), Roberto Trejo-Ayala (6), Karla Salinas-Barboza (7), Yamil Matuk (8), Nadia A. Gandarilla-Martínez (9), Dante Oropeza (10), Sarael Alcauter (3), Miguel E. Rentería (11,12), Alejandra E. Ruiz-Contreras (2*), Alejandra Medina-Rivera (1), Paula Reyes-Pérez (1).
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Abstract:
PALABRAS CLAVE: Enfermedad de Parkinson, Hitos reproductivos, Síntomas no motores, Salud de la mujer, Cuestionario de Salud de la Mujer. INTRODUCCIÓN. La enfermedad de Parkinson (EP) es un trastorno neurodegenerativo progresivo con síntomas motores y no motores. En las mujeres, los factores reproductivos y hormonales, incluidos los efectos neuroprotectores de los estrógenos, pueden influir en el riesgo y la edad de inicio de la EP. Este estudio explora la relación entre los eventos reproductivos y el inicio de la EP en mujeres mexicanas para identificar perfiles de riesgo y orientar enfoques clínicos personalizados. MÉTODOS. Este estudio utilizó datos de la Red Mexicana de Investigación en Parkinson (MEX-PD), con reclutamiento de personas con Parkinson y controles en clínicas neurológicas de todo México. La información reproductiva y hormonal se obtuvo mediante el Cuestionario de Salud de la Mujer (WHQ), y la evaluación clínica incluyó las escalas de Hoehn y Yahr, MoCA y la sección III de la UPDRS. RESULTADOS. Se incluyeron 65 mujeres con EP de la cohorte Mex-PD que completaron el WHQ (Tabla 1). La mayoría se encontraba en etapas tempranas a moderadas de la enfermedad (Hoehn y Yahr mediana = 2; UPDRS III = 33.03 ± 27.42), siendo el estreñimiento (53.8%) y la ansiedad (50.7%) los síntomas no motores más frecuentes. El inicio de la EP ocurrió principalmente en la etapa posmenopáusica (71%), seguido de la premenopausia (16%) y la perimenopausia (12%). Se observaron correlaciones positivas,

aunque no significativas, entre la edad de inicio de la EP y la edad de menarquia, la edad de menopausia y la duración de la menstruación, lo que sugiere la necesidad de estudios con muestras más amplias. CONCLUSIÓN. Estos hallazgos preliminares sugieren que los eventos reproductivos podrían influir en la edad de inicio de la EP en mujeres mexicanas y refuerzan la importancia de integrar la salud reproductiva en el abordaje clínico de la EP.

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