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Abstracts

Universidad Alfonso X el Sabio

Comunicaciones orales

C.1. Modelos de infección por Streptococcus pneumoniae: desde modelos in vitro clásicos hasta minipulmones 3D derivados de hESC

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Streptococcus pneumoniae, o neumococo, es uno de los patógenos responsables de las mayores tasas de morbilidad y mortalidad a nivel mundial. Este microorganismo coloniza de forma asintomática la nasofaringe, pero puede diseminarse hacia otras localizaciones anatómicas, provocando infecciones locales como otitis media o neumonía no bacteriémica, o bien invadir tejidos normalmente estériles, causando bacteriemia o meningitis, lo que se conoce como enfermedad neumocócica invasiva (ENI).

Para el estudio de las infecciones por neumococo, se emplean tanto métodos fenotípicos clásicos, como el serotipado o la determinación de resistencias antibióticas mediante concentración mínima inhibitoria (CMI), como herramientas genotípicas más avanzadas, como la secuenciación del genoma completo. Estos métodos nos permiten evaluar con mayor precisión el impacto de intervenciones de salud pública, como la vacunación.

Más allá del estudio del neumococo en su forma planctónica (de vida libre), es crucial considerar su modo de vida sésil, en forma de biofilm, especialmente en el contexto de interacciones polimicrobianas. En biofilm, la bacteria muestra una mayor resistencia a los tratamientos antibióticos, lo que hace necesario el desarrollo de nuevas estrategias terapéuticas, como el uso de nanomedicamentos.

Finalmente, para investigar de forma más compleja la interacción de neumococo con el hospedador, pueden utilizarse modelos celulares simples, como neutrófilos diferenciados (HL-60 con DMSO) o células epiteliales pulmonares (A549), así como modelos más sofisticados, como mini-organoides pulmonares derivados de células madre embrionarias humanas (hESC), que reproducen de forma más fiel la estructura y función del alveolo pulmonar.

C.2. Impacto del Nicho Hematopoyético en la Transformación del Síndrome Mielodisplásico a Leucemia Mieloide Aguda.

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El Síndrome Mielodisplásico (SMD) es una neoplasia hematológica caracterizada por la transformación maligna de progenitores mieloides provocando una hematopoyesis ineficaz con citopenias periféricas. Aproximadamente el 30-40% de los pacientes con SMD van a evolucionar a Leucemia Mieloide Aguda (LMA) disminuyendo de forma significativa la supervivencia global de éstos. La presencia de mutaciones en los genes ASXL1 o TP53 junto con algunos parámetros clínicos son factores de riesgo para la transformación a LMA. Las Células Estromales Mesenquimales (CEM), son uno de los componentes celulares principales que conforman el nicho hematopoyético que sustenta a los progenitores hematopoyéticos dentro de la médula ósea. Su influencia en la leucemogénesis no está bien estudiada. En este trabajo nos propusimos estudiar si las CEM tenían algún papel en la transformación de los SMD a LMA. Para ello hemos estudiados 49 pacientes con SMD, de los cuales, 29 evolucionaron a LMA y 20 no evolucionaron. Las CEM fueron caracterizadas por citometría de flujo, en diferentes estadíos de la enfermedad. Se identificó una población celular no hematopoyética con alta expresión de CD13, enriquecida con los marcadores canónicos de CEM CD105 y CD90 en el 80% de los pacientes al momento del diagnóstico. El contenido elevado de células CEM CD13 brillantes se asoció significativamente con una progresión más temprana a LMA y una menor supervivencia global. El análisis multivariante confirmó que el contenido de CEM es un predictor independiente de transformación a LMA en pacientes con SMD, lo que resalta su potencial como biomarcador.

C.3. Papel de los fibroblastos asociados a tumores de cáncer de colon.

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El microambiente tumoral, compuesto por células normales, matriz extracelular, vasos sanguíneos y moléculas solubles, juega un papel clave en el desarrollo y progresión del cáncer. En este entorno, los fibroblastos asociados a cáncer (CAFs) destacan como la población celular más abundante, promoviendo la tumorogénesis mediante una intensa comunicación con las células tumorales y otros componentes del microambiente. Entre los principales mediadores de esta comunicación se encuentran los exosomas, que

transportan material genético y proteico entre células locales y distales, contribuyendo al establecimiento de metástasis.
Nuestro grupo se centra en la identificación de nuevos biomarcadores diagnósticos, pronósticos y/o predictivos de respuesta en pacientes con cáncer de colon, con especial atención al papel de los CAFs y los exosomas derivados de estas células. Para ello, y utilizando técnicas de alto interés científico como es el establecimiento de cultivos primarios de CAFs y fibroblastos normales, abordamos distintos objetivos: el análisis de exosomas derivados de estos cultivos, el estudio de la interacción CAF-respuesta a radioterapia estereotáctica (SBRT), así como las interacciones entre microbioma y fibroblastos. La traslabilidad de nuestros resultados la implementamos mediante la evaluación del contenido de los exosomas en biopsias líquidas de pacientes con cáncer de colon y el desarrollo de modelos matemáticos predictivos basados en biomarcadores moleculares e imágenes de TC.
Nuestra investigación tiene un enfoque traslacional y multidisciplinar, en estrecha colaboración con equipos clínicos, garantizando la relevancia clínica de los hallazgos y su potencial aplicación en el contexto de la medicina personalizada en cáncer de colon

C.4. Cómo tratar la obesidad con Neurociencia.

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C.5. The Immune System Paradox: From Guardians to Cancer's Silent Helpers

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The immune system has traditionally been seen as the body's main protector, playing a crucial role in defending us against infections and cancer. However, recent research has challenged this traditional view, revealing that the immune system's role in tumor progression is far more complex and ambiguous than previously thought, contributing to tumor evasion, progression, and dissemination. This talk will provide a theoretical perspective that aims to integrate new insights into the interaction between the immune system and cancer, where beyond being a mere guardian, it is an integral part of a complex ecosystem, with a delicate balance between protection and harm. The

implications of this relationship will also be explored in the development of more effective immunotherapies through the analysis of the mechanisms involved in tumor microenvironment modulation. Finally, the effectiveness of the current strategies will be questioned, suggesting that a deeper understanding of this complex relationship could change the way we approach this disease.

C.6. Una apolipoproteína L humana con actividad similar a un detergente acaba con patógenos intracelulares.

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Intracellular pathogens such as Salmonella enterica serovar Typhimurium (Stm) utilize the host cell cytosol to avoid immune recognition and promote replication. This research highlights Apolipoprotein L3 (APOL3) as a key player in the innate immune defense against cytosolic Gram-negative bacteria. A genome-wide CRISPR-Cas9 screen in human epithelial cells activated by IFN-γ revealed that APOL3 limits intracellular Salmonella replication.

APOL3 functions by specifically interacting with bacterial membranes through a distinct membrane-lytic process. Following IFN-γ activation, APOL3 moves to the cytosol, where it specifically attaches to bacterial membranes that are abundant in acidic phospholipids, causing them to dissolve into nanodiscs and ultimately leading to bacteriolysis. It is supported by other proteins, encoded by IFN-γ stimulated genes (ISGs), that disrupt the outer bacterial membrane, increasing its permeability to APOL3. This mechanism differs from other antimicrobial proteins, emphasizing APOL3's unique function in intracellular protection. Loss-of-function investigations showed that a lack of APOL3 increases bacterial replication in human cells, whereas overexpression diminishes bacterial survival.

These results confirm APOL3 as a strong, IFN-γ-inducible antimicrobial protein that attacks and eliminates Gram-negative bacteria in the cytosol. APOL3 signifies a new pathway in host defense, presenting opportunities for therapeutic use against intracellular bacterial infections.

C.7. Mitochondrial Involvement in Vascular Wall Cells and Its Impact on Atherosclerosis.

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Cardiovascular diseases (CVDs) are the leading cause of death worldwide, with an estimated 17.9 million deaths per year, according to the WHO. Ischemic heart disease generally refers to conditions involving the narrowing or blockage of blood vessels, caused by damage to the heart or blood vessels due to atherosclerosis. Atherosclerosis leads to the hardening and narrowing of arterial walls, which can restrict blood flow through the arteries to organs and tissues, potentially resulting in a heart attack, chest pain (angina), or stroke. For this reason, studying these diseases is essential, as they severely impact health and life expectancy.
Mitochondria play a crucial role in maintaining cellular homeostasis and are highly relevant in the development of cardiovascular diseases, as they are intrinsically involved in the senescence process, during which cells lose their replicative capacity and undergo changes in phenotype and metabolism. These senescent cells remain active and release harmful substances that trigger inflammation and cause damage to neighbouring cells, linking them to cellular aging. Mitochondrial transcription factor A (TFAM) is strongly associated with mitochondrial biogenesis and function maintenance, as well as other mitochondrial and metabolic genes. Therefore, conducting in vitro studies is necessary to understand the mechanisms through which these genes influence mitochondria and their role in atherosclerosis, ultimately leading to more effective diagnostics and treatments.
Keywords: Atherosclerosis, mitochondrial dysfunction, cardiovascular diseases, endothelial dysfunction.

C.8. Memoria.

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Memory is defined as the ability to codify, store and recover information.Considering the amount of time, the information that can be stored and the type of stimuli recorded, there are three types of memory: short-term, long-term and sensory memory. Memory is processed through the hypothalamus and the amygdala. We need to differentiate codification, defined as the first generation of the representation of the information after a stimuli, and consolidation, referred to as the maintenance of an activity pattern.

Processing and retention of the information requires microcircuits that allow to separate information’s patterns. This patterns’ repetition leads to long-term memory consolidation.

Neuronal plasticity is also important in the matter, favouring prolonged synaptic activations. Different types of memory have differential molecule pathways, differing in neurotransmitters and transcription factor activation.

The modulation of memory through different substances such as neurotransmitters and hormones for memory stimulation is also mentioned. This modulation is regulated by the basolateral amygdala and the interaction of these substances with different receptors. We concluded that molecules that interact with noradrenaline can lead to either an increase or decrease of memory capability

C.9. The future of Cancer Immunotherapy: CARBased strategies for solid tumors.

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CAR-T cell therapy has revolutionized cancer treatment, particularly in hematological malignancies. However, its e@icacy in solid tumors has been limited by tumor heterogeneity and the immunosuppressive tumor microenvironment. In response, alternative CAR-based therapies utilizing “non-T cells”, such as CAR-NK (natural killer cells) and CAR-M (modified macrophages), are emerging as promising strategies to overcome these challenges.

This presentation will explore the development and therapeutic potential of CAR-T, CAR-NK, and CAR-M cells in the treatment of solid tumors. We will discuss the distinct biological features and functional capabilities of each cell type, and how they can be engineered to improve tumor infiltration, overcome immune suppression, and target resistant tumor cells. Additionally, recent advancements, ongoing challenges, and the future potential of these therapies will be reviewed, including their combination with other treatments to enhance e@ectiveness.

The goal of this presentation is to provide a comprehensive overview of the evolving landscape of CAR therapies for solid tumors, emphasizing innovative approaches that could significantly impact the future of cancer treatment.

Posters

P.1. Las drogadicciones a opiáceos: un punto de vista psicológico, fisiológico y farmacológico.

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Drug addiction represents a complex and multifactorial issue that can be approached frommultiple disciplines. This work examines addictions; particularly to opioids, from a psychological,neurophysiological, and pharmacological perspective. Fundamental concepts related toaddiction—such as tolerance, dependence, and withdrawal—are discussed and defined by saiddisciplines.

From a neurophysiological standpoint, we will be examining the brain areas, systems, and substances involved in addiction, with detailed emphasis on their effects on dopaminergicpathways. From a pharmacological perspective, we will analyze a general classification of drugsprovided, with a focus on opioids such as fentanyl.

Finally, various psychological guidelines are presented to help identify and treat individuals withdrug addiction in clinical settings.

P.2. Preventing severe allergic reactions with nanoparticles.

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Nowadays, it is not unusual to experience severe allergic reactions (anaphylaxis) to various substances, such as certain foods, antibiotics or insect stings. These allergens are triggered as an immune system response due to the production of IgE and their consequent union to mast cells, causing the release of their granules and can be severe enough to cause death if not treated in time.

That is why, after searching for ways to prevent anaphylaxis, it has been observed that some mast cell proteins can be intercepted to prevent their activation and, therefore, the development of anaphylaxis. For this purpose, nanoparticles made of a material that does not alter the antibodies that recognize these proteins were developed, so that the nanoparticles could be coated with them. After testing and using these nanoparticles, it was found that they can precisely reach these mast cell targets and deliver antibodies to them, which is a breakthrough in curbing severe allergic symptoms.

P.3. AI in biomedicine.

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INTRODUCTION
Artificial intelligence has transformed key industries and, if managed properly, will continue to advance knowledge and solve complex problems, although it faces challenges in regulation, privacy, and ethics.

RELATION BETWEEN AI AND BIOMEDICINE: Automatic diagnosis, Personalized medicine and Drug research
Examples:
- OMS: studies on the use of AI in the detection of diseases such as tuberculosis using machine learning algorithms.
- NIH: Has funded research on AI applied to personalized medicine and drug discovery.
- The Lancet & JAMA (Journal of the American Medical Association): studies on the accuracy of AI algorithms in radiology and dermatology.

*Challenge: AI as a complement, not replacement

AI AND THE HEALTHY AGING: Assistive robots, Early detection of neurodegenerative diseases, Prevention of loneliness
Examples:
- OMS: the use of AI in digital health for older adults and its role in the early detection of Alzheimer's.
- Nature Medicine: prediction neurodegenerative diseases based on speech and movement patterns.
- NEJM (New England Journal of Medicine): assistive robots and their impact on the quality of life of the elderly.
*Challenge: dehumanization
ETHICAL CONSIDERATIONS: Medical automation, Genomics (gene manipulation), Privacy and security of data
Examples:
- OMS: ethical principles for the use of AI in health, highlighting the need for transparency and equity.
- Harvard Medical School: dilemmas about data privacy and security in medical AI.
- BMJ (British Medical Journal): Discusses the regulation and risks of automating medical decisions.
*Challegne: balance
Genomics example: https://doi.org/10.1016/j.artmed.2024.102962
How AI can predict metabolic risks in obese children, enabling personalized interventions that could reduce comorbidities and costs associated with childhood obesity. dasci.es

P.4. Purificación de célula endotelial y pericito de cerebro en modelo de anafilaxia sistémica en ratón.

Andrea Salvador Campos

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Anaphylaxis is defined as a systemic hypersensitivity reaction that occurs rapidly and can become life-threatening. In most cases, it is triggered by an IgE-mediated allergy. This pathology presents a highly diverse etiology, which mainly depends on age, in most cases in children food allergens are the primary cause, whereas in adults, medications and insect venoms are the most common triggers.

Understanding this reaction is crucial due to its widespread impact on the human body, affecting various systems such as the cardiovascular and nervous systems. Additionally, a wide variety of cells and molecules are involved in the process, with potential implications for neuronal symptoms and the blood-brain barrier.

This study focuses on the isolation and characterization of endothelial cells and pericytes from the brains of anaphylactic mouse models. We optimized a flow cytometry-based isolation protocol using specific antibodies, such as CD13 for the pericytes and CD31 for the endothelial cells and explored the potential for culturing these cells for future biomarker and signaling pathways studies. The research particularly focuses on the cortex due to its structural complexity and its proximity to the blood-brain barrier.

By optimizing the isolation and characterization of endothelial cells and pericytes, this study opens the door to future investigations on the role of the blood-brain barrier in anaphylaxis, expanding this work could help identify new biomarkers and therapeutic targets

P.5. Leukocytes associated pathologies.

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Leukocytes are a key component of the immune system responsible for protecting the body against infections or foreign substances, despite making up for only a small fraction of all the blood cells. They come from the blood marrow and differentiate into specialized cells to do so. However, when the immune system overreacts or fails to function properly, it can lead to significant health issues.
This work reviews different immune disorders, focusing on hypersensitivity reactions, autoimmune diseases, immunodeficiencies, and genetic conditions that affect this leukocyte function. It also discusses both acquired immunodeficiencies, like HIV, or genetic disorders such as chronic granulomatous disease.
Understanding how these immune responses are dysregulated is crucial to acquire more knowledge on how to treat and prevent any dysregulation. While a lot of progress has

been made, there is still need for further research, and hopefully one day fully understand how our immune system works.

P.6. Immunological Differences in Meningioma Patients: Impact on Seizure occurrence.

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Background: Meningiomas are the most common type of intracranial primary tumors, and approximately 30-40% of patients experience seizures. The underlying mechanisms remain unknown, but the increasing amount of new evidence suggests that neuro-inflammation maybe the main reason, particularly the role of cytokines IL-6 and TNF-α.

Methods: We propose a prospective study analyzing the immunological differences in meningioma patients with and without seizures. A cohort of 60 patients with confirmed meningiomas (from grade I to III) will be evaluated. Inflammatory biomarkers (IL-6, TNF-α) will be quantified through ELISA, and neuroinflammation will be assessed using PET-TAC imaging. Seizure frequency will be collected via electroencephalogram (EEG), and quality of life will be measured using the EQ-5D scale.

Expected Results: We hypothesize that patients with seizures will exhibit increased levels of IL-6 and TNF-α, along with microglial activation, contributing to blood-brain barrier disruption and neuronal hyperexcitability. Furthermore, we anticipate that targeted immunotherapy will reduce inflammatory markers and seizure frequency.

Conclusion: Identifying specific immunological mechanisms in meningioma-related epilepsy could provide new therapeutic targets. Particularly the immuno-modulation of IL-6 and TNFα may represent a promising strategy to improve seizure control in these patients.

P.7. The Artificial Inteligent in Biomedicine.

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Artificial Intelligence (AI) is a scientific field focused on creating programs that replicate human cognitive processes such as learning and reasoning. Biomedicine brings together disciplines like biochemistry, molecular biology, and genetics, which are essential to modern medicine.

AI allows for faster and more accurate analyses, enabling early disease detection and optimized treatment plans, ultimately improving health outcomes and quality of life.

In medical diagnostics, AI can analyze images and clinical data more efficiently, helping to detect conditions like cardiovascular diseases and cancer from MRIs and X-rays.

In drug development, AI accelerates the discovery process by analyzing large datasets and identifying suitable chemical compounds more quickly. This enhances drug design and supports the creation of personalized therapies for complex conditions.

AI also plays a key role in genetic data analysis by identifying mutations linked to diseases and recognizing patterns in genomic data—pushing forward personalized medicine.
For patient monitoring, AI-powered devices such as smartwatches and biosensors can detect changes in real time and alert users or healthcare providers promptly.
Several companies are already applying this technology. International examples include IBM Watson Health, Tempus, and PathAI. In Spain, companies like Quibim, Inbrain Neuroelectronics, and AItenea Biotech are leading the way.
Despite its benefits, this growing field also presents ethical challenges. Establishing international regulations and standards is essential to ensure patient safety and responsible AI use in healthcare.

P.8. Estudio de la fragmentación de ADN espermático por Ensayo Cometa.

Jacqueline Estefanía Mazorco Vega

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The precise diagnosis of the male component constitutes one of the greatest challenges in the area of assisted human reproduction. Among the most common and least studied causes of male infertility is DNA damage in mature sperm, particularly involved in both embryonic development and pregnancy, which can lead to implantation problems, repeated miscarriages, or pathologies in the newborn. Currently, routine diagnostic tests such as semen analysis are not particularly useful in clearly determining the origin of the defect. The lack of standardization of the techniques involved, as well as their comparison and evaluation of reliability and viability, constitute another additional problem. Therefore, this study aims to analyze the sensitivity and specificity of sperm DNA integrity diagnosis using the neutral comet assay compared to other alternative methodologies. The clinical implementation of analyses such as this one seeks to provide the scientific community with the necessary tools to achieve a fundamental advance that will increase the success of treatments and help patients with difficulties conceiving.

P.9. Cardiac channel molecular autopsy: consecutive cases of autopsy-negative sudden unexplained death referred for Postmortem genetic testing.

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Sudden cardiac death (SCD) constitutes a highly relevant clinical and forensic challenge, particularlywhen it affects young and apparently healthy individuals. Although conventional autopsy enables theidentification of structural abnormalities in many cases, a significant percentage of deaths areclassified as sudden unexplained death (SUD), in which no conclusive morphological or histological findings are observed.

In this context, molecular autopsy emerges as an essential and complementary diagnostic tool, allowing for the detection of underlying genetic alterations that predispose individuals to malignant arrhythmias. This study specifically focuses on two inherited cardiac channelopathies: long QT syndrome (LQTS) and catecholaminergic polymorphic ventricular tachycardia (CPVT). Throughthe analysis of key genes such as KCNQ1, KCNH2, SCN5A, and RYR2, pathogenic mutations can be identified that severely impair the function of cardiac ion channels, disrupting electrical conduction and triggering potentially fatal arrhythmic events.

Furthermore, strong emphasis is placed on the genetic and clinical evaluation of relatives, as the presence of pathogenic variants does not always result in overt clinical symptoms due to variable penetrance and expressivity. Early identification of asymptomatic carriers facilitates the implementation of personalized preventive strategies, including pharmacological treatments with beta-blockers, recommendations for implantable cardioverter-defibrillators (ICDs), and lifestyle adjustments aimed at reducing risk.

Ultimately, the integration of molecular autopsy with family-centered genetic counseling and screening offers a crucial and proactive approach to preventing SCD and safeguarding individuals at risk, highlighting the importance of precision medicine in forensic and clinical cardiology.

P.10. Biomarcadores en endometriosis: avances hacia un diagnóstico no invasivo.

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Endometriosis is a chronic gynaecological disease affecting a large percentage of women of reproductive age, characterised by the presence of endometrial tissue outside the uterus and associated with pelvic pain, infertility and reduced quality of life. Current diagnosis relies on invasive procedures such as laparoscopy, leading to high costs and considerable diagnostic delay. This paper explores the potential of non-invasive biomarkers to facilitate earlier and more accurate diagnosis. Through a literature review focusing on recent studies, promising protein and genetic biomarkers were identified. These include plasma proteins such as interleukin-6, HSP90 and GPx4, found in serum, and several microRNAs (miR-15b, miR-16 and miR-191) whose expression has been seen in cases of endometriosis. These findings suggest that analysis of multiple biological fluids may improve the diagnostic process. While some combined biomarker models already show high accuracy, their clinical application still requires validation in larger studies. This affirms the need for a multidisciplinary approach that integrates molecular, immunological and functional markers, enabling early and personalised diagnosis.

P.11. Regeneración de médula espinal en ratones a partir de células madre.

Alejandra Puerta, Azahara Gil y Beatriz Silvestre, Eugenia Garrido

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Spinal cord injuries (SCI) cause irreversible motor and sensory impairments, with no effective cure currently available. This study explores the neuroregenerative potential of a novel biocompatible hydrogel derived from echinoderms, combined with reduced graphene oxide (rGO) scaffolds, to promote spinal cord repair in a murine model. The hydrogel serves as a neuronal microenvironment that enhances neural stem cell (NSC) adhesion, differentiation, and survival. It is composed of a three-dimensional extracellular matrix with biopolymers such as chitosan, hyaluronic acid, and gelatin, along with sea urchin-derived calcite crystals and adhesive proteins, which provide structural support and integration with neural tissue. rGO scaffolds, fabricated through freeze-drying and characterized for electrical conductivity, were implanted into rats with complete thoracic spinal cord transection (T9-T10). A controlled magnetic field was applied to induce localized heating of iron oxide nanoparticles within the hydrogel, promoting its controlled dissolution and guiding nanoparticle migration towards NSCs and mesenchymal cells. The neuroregenerative effects were assessed through histological (neuronal markers), electrophysiological (neuronal activity), and behavioral (motor recovery) analyses. Results indicated that the Rgo scaffolds facilitated axonal invasion from brainstem motor nuclei, enhancing mechanical stabilization and uniform axon distribution at the lesion site. Additionally, vascular endothelial growth factor (VEGF) expression changes were observed, suggesting a potential role in spinal cord regeneration. These findings highlight the potential of echinoderm-derived hydrogels and rGO scaffolds as promising biomaterials for spinal cord repair, paving the way for future translational research.

Keywords: Reduced graphene oxide, spinal cord injury, neural scaffolds, neuroregeneration, electrophysiology, Wistar rats, Paracentrotus lividus.

P.12. Hurones transgénicos tratados con CRISPR/CAS9 para investigar la enfermedad de Huntington.

Antonia Arizabaleta, Pablo Badolato, Angela Prado y Chen Felipe Yang

RESUMEN La enfermedad de Huntington (HD) es una patología neurodegenerativa, con un patrón de heredabilidad autosómico dominante, causada por la expansión anómala de repeticiones CAG en el gen HTT. La expansión de tripletes da lugar a una forma mutada y tóxica de la proteína huntingtina (HTT) que tiende a acumularse. Esta alteración provoca una progresiva disfunción y pérdida neuronal generando síntomas motores, cognitivos y psiquiátricos. Hasta la fecha, los tratamientos disponibles son exclusivamente sintomáticos y no modifican el curso de la enfermedad. Este proyecto propone un enfoque terapéutico innovador basado en un sistema de edición génica CRISPR-Cas9 dirigido específicamente a neuronas mediante un promotor neuronal (CaMKIIα). El sistema CRISPR

tiene el objetivo de reducir o corregir la expresión de la HTT mutada in vivo, ya que está diseñado para poder ser administrado en etapas posnatales. Para ello, se desarrollará un modelo transgénico de hurón (mustélido) portador de la mutación en el gen HTT, permitiendo una aproximación traslacional más realista. Se evaluará la eficacia del tratamiento mediante la cuantificación de los niveles de huntingtina, junto con el análisis de alteraciones histológicas y conductuales. La metodología incluye técnicas como microinyección intracraneal de AAV9, diseño de ARN guía específico, RT-qPCR, Western Blot, HTRF e inmunohistoquímica, proporcionando una evaluación integral del impacto terapéutico y desarrollo de la enfermedad. Finalmente, los resultados obtenidos con el sistema CRISPR serán comparados con una estrategia alternativa basada en oligonucleótidos antisentido (ASO), una línea de tratamiento que se muestra prometedora para modificar el curso de la enfermedad.


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