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Ministerio de Salud

Instituto Nacional de Ciencias Neurológicas



ANTOLOGÍA DE ARTÍCULOS EN CIENCIAS NEUROLÓGICAS INCN (2025)



LIMA, JULIO 2026



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ANTOLOGÍA DE ARTÍCULOS EN CIENCIAS NEUROLÓGICAS INCN (2025)

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PRESENTACIÓN

El Instituto Nacional de Ciencias Neurológicas (INCN), entidad desconcentrada del Ministerio de Salud y líder en la atención altamente especializada en neurología y neurocirugía que integra la práctica clínica, investigación, docencia, e innovación tecnológica; promueve la capacitación continua del potencial humano como el pilar fundamental para el desarrollo institucional y la mejora de la calidad en la atención especializada, que se extiende al fortalecimiento de la difusión del conocimiento científico.

La Oficina Ejecutiva de Apoyo a la Investigación y Docencia Especializada (OEAIDE) del INCN, constituye el eje estratégico que integra la generación, el fortalecimiento y la difusión del saber neurológico, a través de la Unidad de Investigación que dirige, planifica y supervisa la investigación promoviendo la publicación de artículos científicos en salud neurológica; la Unidad de Docencia que asegura la capacitación continua, la articulación entre la formación académica y práctica clínica, y promueve la gestión del conocimiento científico y servicios bibliotecarios para la optimización del potencial humano; la Unidad de Publicaciones e Innovación Tecnológica, que materializa estos esfuerzos garantizando que la producción institucional trascienda al ámbito académico nacional e internacional.

La OEAIDE, en virtud de la democratización del acceso a la información científica, ha elaborado, a través de la Biblioteca Institucional, la publicación “Antología de Artículos en Ciencias Neurológicas INCN (2025)”, la cual forma parte de la Serie de Publicaciones Bibliográficas y Científicas N°3 (SPByC), y tiene por objetivo central difundir nuevos conocimientos y organizar la información especializada generada por la institución en beneficio de la comunidad científica global, con énfasis en ciencias neurológicas.

Dicha publicación recopila 51 artículos científicos acreditados e indexados en revistas nacionales e internacionales que incluyen la participación de destacados autores afiliados al INCN, su estructura comprende: título, tipo de artículo, autor/es y filiaciones, cita de la fuente, resumen, carátula del artículo y enlace al texto completo. Asimismo, los contenidos abarcan áreas críticas de la neurociencia contemporánea, destacando los temas de: enfermedades neurodegenerativas, neuroinfecciones y parasitosis del sistema nervioso, enfermedades cerebrovasculares y neurocirugía, neurogenética y enfermedades raras, bioética, educación y gestión de la salud, y otras patologías.

En mi calidad de director ejecutivo de la OEAIDE-INCN, es de gran satisfacción poner al alcance de los profesionales, investigadores y académicos de la salud este valioso número de la SPByC; con la publicación de estos trabajos reafirmamos nuestro compromiso con la excelencia académica y la difusión científica, con la convicción de que esta obra fortalecerá el conocimiento y la práctica clínica en nuestro país.

Segundo Carlos Abanto Argomedo

Director Ejecutivo de Apoyo a la Investigación y
Docencia Especializada

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ANTOLOGÍA DE ARTÍCULOS EN CIENCIAS NEUROLÓGICAS INCN (2025)

Poster presentation

1. A novel stop-gain SORL1 mutation from Amerindian background in a Peruvian family with Alzheimer's Disease of the PeADI Study

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Alzheimer's & Dementia, 2025 Jan 3;20(Suppl 1)

Abstract

Background

Common and rare variants in *SORL1* have been associated with increased risk of Alzheimer's disease (AD). Since 2019, we have run an international collaborative research initiative to ascertain a Peruvian cohort for Alzheimer's disease and other related dementias for genetic studies (PeADI).

Method

A Peruvian family (4 AD cases and two mild cognitive impairment (MCI) cases) was recruited through the PeADI study. All six family-member completed a full cognitive assessment, had plasma-based biomarkers pTau181 and Aβ42/40 measured via SIMOA chemistry on the Quanterix HD-X, and underwent whole genome sequencing. Variants within AD risk genes as determined by the ADSP Gene Verification Committee were prioritized and variant interpretation was performed according to ACMG recommendations.

Result

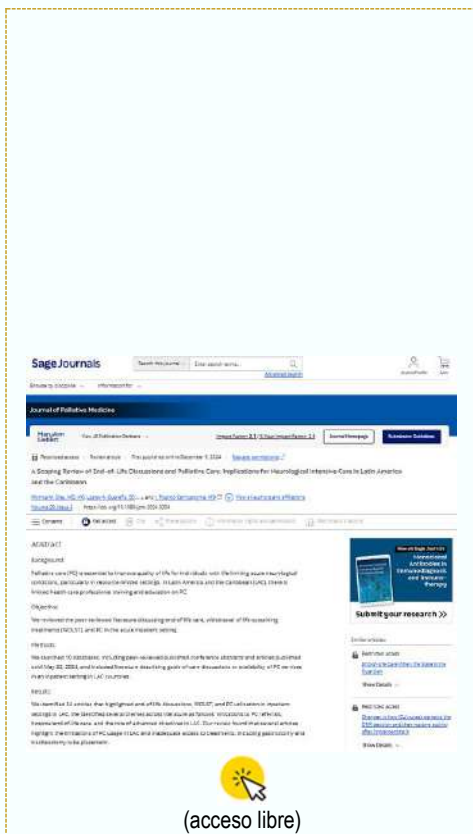
We identified a *SORL1* c.5019G>A (p.Trp1673Ter) variant of Amerindian background in the four AD diagnosed siblings within this family. The two MCI cases did not carry the novel variant. The identified *SORL1* variant corresponded to a heterozygous stop-gain variant in exon 36 replacing tryptophan by a stop codon at position 1673 of the *SORL1* protein. In-silico analysis predicts this variant promotes nonsense-mediated mRNA decay. This variant has not been previously reported in databases including gnomAD, LOVD and ClinVar. The 4 AD cases had on average 2.3X higher plasma pTau181 concentrations compared to the 2 MCI (2.03 ± 0.28pg/μl vs 0.88 ± 0.7pg/μl). There was no noticeable difference in the Aβ42/40 ratio. This variant is classified as likely pathogenic according to ACMG.

Conclusion

We report the first Peruvian AD family carrying a likely pathogenic stop-gain *SORL1* variant within an Amerindian background region. Further cosegregation and functional assays are required to establish the risk size of this variant for AD.



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Review article

2. A Scoping Review of End-of-Life Discussions and Palliative Care: Implications for Neurological Intensive Care in Latin America and the Caribbean

Monica M. Diaz, Lesley A. Guareña, Bettie Garcia, Christopher A. Alarcon-Ruiz, Stella M. Seal, Clio Rubinos, Dulce Cruz-Oliver, and J. Ricardo Carhuapoma / [View all authors and affiliations](#)

Journal of Palliative Medicine, 2024

Background

Palliative care (PC) is essential to improve quality of life for individuals with life-limiting acute neurological conditions, particularly in resource-limited settings. In Latin America and the Caribbean (LAC), there is limited health care professional training and education on PC.

Objective

We reviewed the peer-reviewed literature discussing end-of-life care, withdrawal of life-sustaining treatments (WOLST), and PC in the acute inpatient setting.

Methods

We searched 10 databases, including peer-reviewed published conference abstracts and articles published until May 22, 2024, and included literature describing goals-of-care discussions or availability of PC services in an inpatient setting in LAC countries.

Results

We identified 34 articles that highlighted end-of-life discussions, WOLST, and PC utilization in inpatient settings in LAC. We identified several themes across literature as follows: limitations to PC referrals, hospice/end-of-life care, and the role of advanced directives in LAC. Our review found that several articles highlight the limitations of PC usage in LAC and inadequate access to treatments, including gastrostomy and tracheostomy tube placement.

Conclusions

Our review demonstrates a need to improve PC knowledge and access to end-of-life care resources. Regional educational efforts are needed to improve PC knowledge among health care providers who care for patients with acute neurological conditions in LAC.



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Case report

3. Acute cholecystitis in H-type duplicated gallbladder: a case report

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Journal of Surgical Case Reports, Volume 2025, Issue 9, September 2025

Abstract

Gallbladder duplication is a rare congenital anomaly. Abnormal biliary anatomy is associated with an increased risk of complications, such as bile duct injury, during cholecystectomy. In this article, we present a clinical case of gallbladder duplication identified preoperatively by magnetic resonance cholangiopancreatography, which guided surgical planning. A 70-year-old man

was admitted with acute cholecystitis, low-grade fever, jaundice, and dyspnea. Magnetic resonance cholangiopancreatography revealed a duplicated gallbladder (Type H). During surgery, the chronically inflamed upper gallbladder was completely resected. The larger lower gallbladder, with acute cholecystitis and a Mirizzi-like pattern, was partially resected due to severe inflammation and the inability to identify the cystic duct. The patient had an uneventful course. Gallbladder duplication is a rare congenital anomaly that may be associated with other congenital anomalies. Thorough preoperative imaging studies, meticulous surgical technique, and rigorous intraoperative monitoring are essential, as these abnormalities can lead to serious injuries.

Addendum de artículo

4. Addendum: Evidence for transmission of *Taenia solium* taeniasis/cysticercosis in a rural area of Northern Rwanda

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Front. Vet. Sci., 2025

This article is an addendum to:

Evidence for Transmission of *Taenia solium* Taeniasis/Cysticercosis in a Rural Area of Northern Rwanda

Read original article: [Frontiers | Evidence for Transmission of Taenia solium Taeniasis/Cysticercosis in a Rural Area of Northern Rwanda](#)

Research article

5. Ancestral genomic functional differences in oligodendroglia: implications for Alzheimer's disease

Aura M. Ramirez¹ Luciana Bertholim Nasciben¹ Sofia Moura¹ Lauren Coombs¹ Farid Rajabli^{1,2} Brooke A. DeRosa¹ Patrice L. Whitehead¹ Larry D. Adams¹ Takiyah D. Starks³ Pedro R. Mena¹ Maryenela Illanes-Manrique^{4,5} Sergio Tejada¹ Goldie S. Byrd³ Allison Caban-Holt³ Michael L. Cuccaro^{1,2} Katalina McInerney^{1,2} Mario R. Cornejo-Olivas^{4,5} Briseida E. Feliciano-Astacio⁶ Liyong Wang^{1,2} Maria C. Robayo¹ Wanying Xu⁷ Fulai Jin⁷ Margaret A. Pericak-Vance^{1,2} Anthony J. Griswold^{1,2} Juan I. Young^{1,2} Derek M. Dykxhoorn^{1,2} Jeffery M. Vance^{1,2}.

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Alzheimer's & Dementia, Volume 21, Issue 9, September 2025

Abstract

Introduction

This study investigates ancestry-specific changes in induced pluripotent stem cell (iPSC)-derived oligodendroglia genomic regulation in Alzheimer's disease



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(AD), addressing diversity gaps by including African, Amerindian, and European ancestries in the analysis.

Methods

We generated 12 iPSC lines from AD patients and controls with different apolipoprotein E (*APOE*) genotypes, *APOE* $\epsilon 3/\epsilon 3$ and *APOE* $\epsilon 4/\epsilon 4$, across three ancestries. Lines were differentiated into neural spheroids containing oligodendrocyte lineage cells and analyzed by single-nucleus RNA sequencing, Assay for Transposase-Accessible Chromatin with sequencing (ATACseq)APO, and High-throughput Chromosome Conformation Capture (Hi-C).

Results

We identified ancestry-specific differences in gene expression and chromatin accessibility of AD genome-wide association study candidate genes. *APOE* $\epsilon 4/\epsilon 4$ carriers across all ancestries showed upregulated cholesterol biosynthesis genes with decreased myelination markers. iPSC-derived oligodendrocytes demonstrated high correlation ($R^2 > 0.85$) with human brain transcriptomes.

Discussion

Our findings highlight the importance of studying diverse ancestries in AD research and suggest early *APOE* $\epsilon 4$ effects on cholesterol metabolism. The validated iPSC model provides a valuable tool for investigating ancestry-specific disease mechanisms.

Poster presentation

6. Ancestry-specific studies on Alzheimer's disease using iPSC-derived microglia

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Alzheimer's & Dementia, Vol. 20 (december 2024), 2025

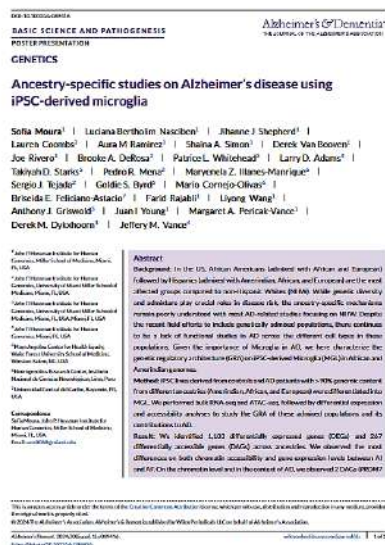
Abstract

Background

In the US, African Americans (admixed with African and European) followed by Hispanics (admixed with Amerindian, African, and European) are the most affected groups compared to non-Hispanic Whites (NHW). While genetic diversity and admixture play crucial roles in disease risk, the ancestry-specific mechanisms remain poorly understood with most AD-related studies focusing on NHW. Despite the recent field efforts to include genetically admixed populations, there continues to be a lack of functional studies in AD across the different cell types in these populations. Given the importance of Microglia in AD, we here characterize the genetic regulatory architecture (GRA) on iPSC-derived Microglia (MGL) in African and Amerindian genomes.

Method

iPSC lines derived from controls and AD patients with >90% genomic content from different ancestries (Amerindian, African, and European) were differentiated into MGL. We performed bulk RNA-seq and ATAC-seq, followed by differential expression and accessibility analyses to study the GRA of these admixed populations and its contributions to AD.



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Result

We identified 1,103 differentially expressed genes (DEGs) and 267 differentially accessible genes (DAGs) across ancestries. We observed the most differences on both chromatin accessibility and gene expression levels between AI and AF. On the chromatin level and in the context of AD, we observed 2 DAGs (*PRDM7* and *SCIMP*) between AI and AF, and 1 DAG between AI and EU (*PRDM7*). In addition, we identified 10 AD-risk modifying genes that are differentially expressed between AI and AF ancestries (*ABI3*, *CTSB*, *JAZF1*, *MS4A6A*, *PILRA*, *PLEKHA1*, *RASGEF1C*, *SORL1*, *TREM2*, and *TREML2*) and 3 DEGs between AI and EU (*JAZF1*, *MS4A6A*, and *SORL1*). We identified several DEGs to be involved in lipid metabolism, cholesterol biosynthesis and metabolism, lysosomal activity, and immune response - all highly relevant processes in AD pathology.

Conclusion

We provide new insights into ancestry-specific genetic risk factors in AD pathophysiology. Here, we report novel transcriptomic and chromatin accessibility data in microglia of AI and AF ancestries that potentially contribute to a differential genetic risk in AD in the different ancestries. Interestingly, those ancestries with greatest migratory differences revealed the largest DEG.

Research article

7. APOE and Alzheimer's disease and related dementias risk among 12,221 Hispanics/Latinos

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Alzheimer's Dementia, 2025

Abstract

Background

Effect of apolipoprotein E (*APOE*) on Alzheimer's disease and related dementias (ADRD) risk is heterogeneous across populations, with scarce data on Hispanics/Latinos.

Methods

APOE genotype was studied in 12,221 Hispanics/Latinos (per cohort and via metanalysis): Caribbean-Hispanics, Mexicans, Mexican-Americans, and



Peruvians/Bolivians. A subsample had longitudinal assessment and plasma p-tau. We tested the modifying effects of global and local ancestries. Results were replicated in an independent Peruvian cohort and brain samples.

Results

APOE $\epsilon 4$ effect was strongest in Peruvians/Bolivians (odds ratio [OR] = 6.13, 95% confidence interval [CI] = 2.71–13.83), followed by Mexicans (OR = 4.31, 95% CI = 1.58–11.74), Mexican-Americans (OR = 3.06, 95% CI = 2.04–4.59), and Caribbean-Hispanics (OR = 2.22, 95% CI = 1.99–2.48). Meta-analyses showed OR = 2.32 (95% CI = 2.09–2.57) and OR = 0.81 (95% CI = 0.68–0.97) for the $\epsilon 4$ and $\epsilon 2$ allele, respectively. The *APOE* $\epsilon 4$ effect was replicated independently in Peruvians (OR = 5.06, 95% CI = 2.48–10.70). $\epsilon 4$ carriers displayed higher AD/AD risk, and p-tau levels. Global and local ancestries did not modify AD/AD risk, and they were associated with Braak stage.

Discussion

APOE shows a heterogeneous effect on AD/AD risk in our Hispanics/Latinos sample, the largest to date.

Poster presentation

8. Attitudes and Perceptions of Caregivers of person with Frontotemporal Dementia in Peru: Preliminary results

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Alzheimer's & Dementia, 2025 Jan 9;20(Suppl 4)

Abstract

Background

Frontotemporal dementia (FTD) is a clinical syndrome characterized by progressive changes in behavior, executive function, or language. In Latin America, persons with FTD are underrecognized or diagnosed late. There is a lack of knowledge about the experiences have on families

Method

We used a mixed methods approach consisting of qualitative, semi-structured interviews that were recorded, transcribed, and analyzed using thematic analysis. Local IRB approval was obtained for this study.

Result

We interviewed fourteen family caregivers of persons with FTD, ten of them (71.4%) were caregivers of persons with behavioral variant FTD (bvFTD). There was female predominance (78.5%). The average of education was 14.5±2.3 years. Half were sole caregivers, seven were children, five were spouses, and two were sisters. The key themes emerging from qualitative interviews were difficulties surrounding the diagnostic process, the impact of knowing the diagnosis, and challenges surrounding caregiving. Caregivers stressed how tedious it is to access health services and obtain specialized care and the lack of knowledge of the disease among health professionals. Caregivers commented on the perceived lack of skills of some professionals when informing about the diagnosis and prognosis of a neurodegenerative disease. Knowledge of FTD prognosis generates anguish, hopelessness, and frustration among caregivers. Caregivers who are sons or daughters often adopt leadership roles within the family, especially from financial and caregiving point of view. They also often change or postpone life/professional plans. On the other hand, if the caregiver is the wife or husband, the absence of their partner in the home stands out as a main stressor, even though they remain together.



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Conclusion

Our preliminary results highlight the importance of providing accessible, plain-language education and support to caregivers of persons with FTD, as well as the opportunity to improve training on healthcare professionals. The impact of caring for patients with FTD appears to be dependent on the parental relationship.

Review article

9. Calcified Neurocysticercosis: Understanding Dead (Not Necessarily Inactive) Parasites

Christina M. Coyle, Javier A. Bustos, Oscar H. Del Brutto, Hector H. Garcia, Theodore E. Nash, Vedantam Rajshekhar

The American Journal of Tropical Medicine and Hygiene, Volume 112, Issue 6, 2025

Abstract

Neurocysticercosis (NCC) is the most common helminthic infection of the human central nervous system (CNS), and a major cause of acquired epilepsy in most of the world. Although outcomes of NCC have been improved by advanced diagnostic tests, antiparasitic drugs, and appropriate anti-inflammatory treatment, little attention has been paid to calcified NCC. Calcification is a common outcome in NCC and carries increased risk for seizures and hippocampal atrophy/sclerosis. The pathophysiological mechanisms leading to calcification instead of complete resolution are basically unknown. Understanding the causes and mechanisms of calcification can lead to improved therapies aimed at reducing the likelihood of residual calcification or the underlying pathological mechanisms, after the resolution of parasitic lesions in the human CNS.




(suscripción o compra)

Poster presentation

10. Clinical profile of Amnestic Early-Onset Alzheimer's disease in Peru: Case Series from a Neurological Care Center

Sheila Castro-Suarez^{1,2,✉}, Erik Alberto Guevara-Silva¹, Kelvin Alvarez-Toledo¹, Cesar Caparo-Zamalloa¹, Jonathan Adrian Zegarra-Valdivia³, Maria Meza-Vega^{1,4}

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Alzheimer's & Dementia, 2025 Jan 3;20(Suppl 3)

Abstract

Background

Early-onset Alzheimer's disease (EOAD) is a rare and devastating form of Alzheimer disease that represents 5-10% of the total number of Alzheimer disease (AD). It is significantly less well studied than the late-onset form of AD. The clinical presentation is heterogeneous, the amnestic variant is the most frequent (75%). Clinical aspects of EOAD in Admixed Latin American population are lacking in the literature. We aim to describe clinical features of amnestic EOAD patients followed-up at a Neurological healthcare center in Peru.

Method

We reviewed medical records of patients with diagnosis of probable EOAD during January-December 2023. All patients were evaluated by trained neurologists from a dementia outpatient clinic at a specialized neurologic center in Lima, Peru. Relevant information about neurocognitive, neuropsychiatric, and functional assessments were extracted from medical records with further descriptive analysis. IRB approval from local institution was obtained for this study.




(acceso libre)

Result

A total of 45 patients (73% female, age at onset = 56 [50-61] years) met the selection criteria. The 71% of the cases have more than 6 years of education. The delay for diagnosis in this group was between 2 and 5 years. A first-degree relative with dementia was present in 15.5% of patients. Clinical and cognitive assessment profile included: MMSE average score was 13.2 ± 6.8 . The clinical manifestation characterized by anterograde episodic memory impairment were accompanied by executive disfunction (97.8%), attention and calculation impairment (84.4%), and visuospatial impairment (71%); impaired repetition of phrases and anomia were present in 67 and 44% respectively. Based on NPI the most frequent neuropsychiatric symptoms were depression (57%) irritability (51%), and anxiety (38%). Severity of dementia was CDR = 3 for 31.1%, CDR = 2 for 35.6%, CDR = 1 for 33.3%.

Conclusion

The clinical features of our Peruvian amnesic EOAD cohort are mostly consistent with previous reports. There is a significant delay of EOAD diagnosis. Implementing training strategies for clinicians, neurologists, and neurologists, will improve a timely and accurate diagnosis of EOAD.

Original breve

11. Clinical profile of early-onset Alzheimer's disease in Peru: case series from a neurological care center

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Revista Peruana de Medicina Experimental y Salud Pública, 2025; 42(2)

Resumen

La enfermedad de Alzheimer de inicio precoz (EAIP) representa entre el 5 y 10% de todos los casos de enfermedad de Alzheimer, constituye una forma rara y devastadora de esta condición. Este estudio retrospectivo analizó las historias clínicas de pacientes diagnosticados con EAIP entre 2022 y 2023 en un centro neurológico terciario en Lima, Perú. De 547 casos de demencia, 60 cumplían criterios para EAIP. La mayoría fueron mujeres (73,3%), el 71% tenía más de seis años de educación. La media del MMSE fue de $11,92 \pm 7,5$ y la del CDR fue de 2, indicando deterioro cognitivo severo y demencia moderada. La variante amnésica fue la forma clínica más común, destacando el compromiso de la memoria episódica y la función ejecutiva. Los síntomas psicológicos y conductuales más frecuentes fueron la depresión y la irritabilidad. También se identificaron características atípicas como mioclonías (5%) y epilepsia (10%). Estos hallazgos destacan la importancia de que los profesionales de salud reconozcan la demencia en adultos jóvenes, en fases tempranas y ofrezcan un manejo integral que mejore la calidad de vida de los pacientes y sus familias.



(acceso libre)

Poster presentation

12. Cognitive Impairment and Dementia Prevalence in the Northern Region of Peru: An Epidemiological Overview

Jonathan Adrian Zagarra-Valdivia¹; Leandro Aron Perez-Fernandez¹; Milagros del Rocío Casimiro-Arana¹; Harold Alessandro Arana-Nombero¹; Viviana Nayelli Gallegos-Manayay^{1,2}; Reyna Elizabeth Alamo-Medina¹; María del Rosario Oliva-Piscoya¹; Eduardo Sebastián Abanto-Saldaña¹; María Celinda Cruz Ordinola¹; Nobuko Vásquez-Zuñe¹; Sheila Castro-Suarez^{3,4}; Carmen Paredes Manrique⁵; Lisseth Catherime Detquizan Pérez¹; Ernesto Sebastian Lozano Fernández¹; Leslie Magdalena Lozada Tantarico¹; Zaráid Maria Chancafe Neciosup¹; Mariana Villegas¹; Julio Inga¹; Sintia Vásquez¹; Karolay Lluén Navarrete¹; Erick Muñoz¹; Nilson Domínguez¹; Julio Ramírez¹; Nataly Diaz¹; Nelson Salinas¹; Johnny Ramos¹; Patricia Ventura¹; Deliz Liza Serrán¹; Josetty Coronado¹.

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Alzheimer's & Dementia, Vol. 20 (december 2024), 2025

Abstract

Background

The prevalence of dementia in Peru's northern regions is poorly documented, largely due to the scarcity of studies employing validated assessment tools for the elderly. Notably, this area is marked by pronounced disparities, encompassing a wide range of socioeconomic statuses and predominantly low educational attainment. The confluence of risk factors, including educational and socioeconomic deprivation, prevalent diseases, suboptimal health conditions, chronic stress, and lifelong malnutrition, poses a significant risk of escalating dementia cases over the next two decades.

Objective

This study aims to ascertain the prevalence of cognitive impairment among a socioeconomically diverse cohort of individuals aged 50 and above in Chiclayo, attending various community settings such as Senior Centers, Nursing Homes, and neighborhood groups. It zeroes in on the northern region, notorious for its pronounced cognitive disparities largely attributed to educational variances.

Methods

Employing a descriptive epidemiological approach with a cross-sectional design, this study utilizes a two-stage cluster sampling methodology. To date, over 400 participants from the northern region have been evaluated. The assessment includes cognitive screenings to pinpoint cognitive deficits and functionality issues, alongside the exclusion of potential emotional disorders.

Results

Initial findings from 99 qualifying participants reveal the Rowland Universal Dementia Assessment Scale - Peru (RUDAS-P) as an effective tool for identifying cognitive impairment and dementia in populations with limited education. Despite varied educational backgrounds, 25% exhibited cognitive impairment, and 16% showed dementia signs, aligning with findings from other national studies. The evaluation of executive functions unveiled significant inter-group variances. However, a universally diminished executive performance score across all participants, including the control group, was observed, likely attributable to an amalgamation of risk factors such as malnutrition and unmanaged chronic conditions like diabetes and hypertension.

Conclusion

This research strives to generate prevalence data on cognitive impairment and dementia among Chiclayo's elderly, mirroring the area's socio-economic diversity. By spotlighting the challenges of pathological aging and prevalent risk factors, the study underscores the critical need for heightened public awareness and proactive brain health maintenance to thwart dementia onset.



(acceso libre)



Research article

13. Comparing Alzheimer's genes in African, European, and Amerindian induced pluripotent stem cell-derived microglia

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Alzheimer's & Dementia, 2025 Feb;21(2)

Abstract

Introduction

Genome-wide association studies (GWAS) studies in Alzheimer's disease (AD) demonstrate ancestry-specific loci. Previous studies in the regulatory architecture have only been conducted in Europeans (EUs), thus studies in additional ancestries are needed. Given the prevalence of AD genes expressed in microglia, we initiated our studies in induced pluripotent stem cell (iPSC) - derived microglia.

Methods

We created iPSC-derived microglia from 13 individuals of either high Amerindian (AI), African (AF), or EU global ancestry, including both AD and controls. RNA-seq, ATAC-seq, and pathway analyses were compared between ancestries in both AD and non-AD genes.

Results

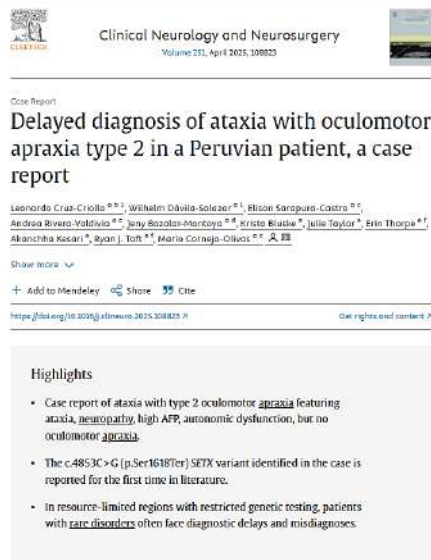
Twelve AD genes were differentially expressed genes (DEGs) and/or accessible between ancestries, including ABI3, CTSB, and MS4A6A. A total of 5% of all genes had differential ancestral expression, but differences in accessibility were less than 1%. The DEGs were enriched in known AD pathways.

Discussion

This resource will be valuable in evaluating AD in admixed populations and other neurological disorders and understanding the AD risk differences between populations.

Highlights

First comparison of the genomics of AI, AF, and EU microglia. Report differences in expression and accessibility of AD genes between ancestries. Ancestral expression differences are greater than differences in accessibility. Good transcriptome correlation was seen between brain and iPSC-derived microglia. Differentially expressed AD genes were in known AD pathways.



(suscripción o compra)

Case report

14. Delayed diagnosis of ataxia with oculomotor apraxia type 2 in a Peruvian patient, a case report

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Clinical Neurology Neurosurgery, 2025**Abstract****Introduction**

Ataxia with oculomotor apraxia type 2 (AOA2) is a rare autosomal recessive cerebellar ataxia characterized by progressive cerebellar ataxia, sensorimotor peripheral neuropathy, and occasional oculomotor apraxia.

Case report

A 50-year-old male with a history of orthopedic shoe use since childhood presented with slowly progressive ataxia and neuropathy. Laboratory tests showed elevated serum alpha-fetoprotein levels and increased total cholesterol. Clinical whole genome sequencing identified a c.4853C > G (p.Ser1618Ter) homozygous pathogenic variant in SETX.

Conclusion

The case highlights the challenges identifying rare disorders like AOA2 due to limited access to genetic testing and socioeconomic and healthcare barriers.

Poster presentation

15. Depression and cardiovascular symptoms in a diverse Frontotemporal Dementia cohort, enriched for Caribbean Hispanic patients

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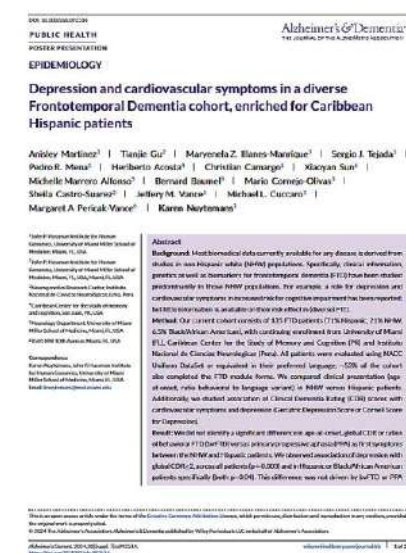
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Alzheimer's & Dementia, Volume 20, Issue S7**Abstract****Background**

Most biomedical data currently available for any disease is derived from studies in non-Hispanic white (NHW) populations. Specifically, clinical information, genetics as well as biomarkers for frontotemporal dementia (FTD) have been studied predominantly in those NHW populations. For example, a role for depression and cardiovascular symptoms in increased risk for cognitive impairment has been reported; but little information is available on their risk effect in (diverse) FTD.

Method

Our current cohort consists of 135 FTD patients (71% Hispanic, 21% NHW, 6.5% Black/African American), with continuing enrollment from University of Miami (FL), Caribbean Center for the Study of Memory and Cognition (PR) and Instituto Nacional de Ciencias Neurológicas (Peru). All patients were evaluated using



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NACC Uniform DataSet or equivalent in their preferred language; ~53% of the cohort also completed the FTD module forms. We compared clinical presentation (age-at-onset, ratio behavioral to language variant) in NHW versus Hispanic patients. Additionally, we studied association of Clinical Dementia Rating (CDR) scores with cardiovascular symptoms and depression (Geriatric Depression Score or Cornell Score for Depression).

Result

We did not identify a significant difference in age-at-onset, global CDR or ratios of behavioral FTD (bvFTD) versus primary progressive aphasia (PPA) as first symptoms between the NHW and Hispanic patients. We observed association of depression with global CDR ≥ 2 , across all patients ($p = 0.003$) and in Hispanic or Black/African American patients specifically (both $p \sim 0.04$). This difference was not driven by bvFTD or PPA subtypes specifically. No significant association was observed for global CDR with diabetes, hypertension or hyperlipidemia in all, Hispanic or NHW patients.

Conclusion

Biomedical research of FTD in underrepresented population groups is necessary as data from research in NHW is not always generalizable across race/ethnicity. Our data suggests that the reported association of depression with higher CDR scores is valid in diverse FTD patient groups; whereas we see less evidence for cardiovascular symptoms being associated with risk for FTD. We are working to expand our efforts characterizing the full clinical and biological presentation of FTD in non-NHW population groups. The biomedical characterization of FTD across race/ethnicity will help the understanding of disease mechanisms in all patients, ultimately preventing further health disparities.

Clinical issues

16. Differences and contributors to global cognitive performance in the underrepresented Latinx Parkinson's disease population

Saar Anis, Henry Mauricio Chaparro-Solano, Thiago Peixoto Leal, Scott A. Sperling, Claire Sonneborn, Camila Callegari Piccinin, Miguel Inca-Martinez, Mario Cornejo-Olivas, Maryenela Illanes-Manrique, Pedro Chana-Cuevas, Paula Safie Awad, Ana Jimena Hernández-Medrano, Amin Cervantes-Arriaga, Artur F. S. Schuh, Carlos R. M. Rieder, Pedro Braga-Neto, Antonio Andrei da Silva Sena, Bruno Lopes Santos-Lobato, Emilia M. Gatto, Griselda J. Alvarado, Cesar L. Avila, Vitor Tumas, Maria Paula Foss, Vanderci Borges, Henrique Ballalai Ferraz, Jorge Luis Orozco Vélez, Beatriz Muñoz Ospina, Sonia Moreno, David Pineda, Julia Esther Rios Pinto, Patricio Olguín, Juan Cristobal Nuñez, Angel Viñuela, Alan O. Espinal-Martinez, Nicanor Mori, Koni Mejía-Rojas, Angel Medina-Colque, Ana Lucia Zuma Rosso, Celmir Vilaca, Edward Ochoa-Valle, Iván Cornejo-Herrera, Paula Reyes-Perez, Alejandra Lázaro-Figueroa, Anna Letícia de Moraes Alves, Rubens Gisbert Cury, Hubert H. Fernandez, Ignacio Mata & The Latin American Research consortium on the Genetics of Parkinson's Disease (LARGE-PD)

The Clinical Neuropsychologist Volume 39, 2025 - Issue 7

Abstract

Objective

Despite significant progress in understanding the factors influencing cognitive function in Parkinson's disease (PD), there is a notable gap in data representation for the Latinx population. This study aims to evaluate the contributors to and disparities in cognitive performance among Latinx patients with PD. **Methods:** A retrospective analysis was conducted based on cross-sectional data encompassing demographic, environmental, motor, and non-motor disease characteristics from the Latin American Research Consortium on the Genetics of PD (LARGE-PD) and the Parkinson's Progression Markers Initiative (PPMI) cohorts. Linear regression multivariable models were applied to identify variables affecting Montreal Cognitive Assessment (MoCA) scores, accounting for age, sex, and years of education. **Results:** The analysis comprised of 3,054 PD patients (2,041 from LARGE-PD and 1,013 from PPMI) and 1,303 Latinx controls. Latinx PD patients (mean age 63.0 $\pm$ 11.8, 56.8% male) exhibited a significantly lower average MoCA score ($p < .001$) compared to white non-Hispanic PD patients from PPMI (mean age 67.3 $\pm$ 9.9, 61.7% male). This difference persisted when comparing the Latinx PD to the Latinx controls (mean age 59.7 $\pm$ 9.3, 53.2% male; $p < .001$). Factors significantly associated with better MoCA scores in Latinx PD included: Unilateral symptom onset ($p < .009$) and higher educational attainment ($p < .001$). Conversely, those associated with worse scores included the use of dopamine agonists ($p = .01$), pramipexole use ($p = .01$), older age ($p < .001$), and a higher Hoehn and Yahr scale score ($p < .001$). **Conclusions:** Latinx PD patients demonstrated significantly lower cognitive scores compared to their white non-Hispanic PD counterparts and Latinx controls. These results highlight the importance of interpreting MoCA scores in a nuanced manner within diverse populations.

Methods

A retrospective analysis was conducted based on cross-sectional data encompassing demographic, environmental, motor, and non-motor disease characteristics from the Latin American Research Consortium on the Genetics of PD (LARGE-PD) and the Parkinson's Progression Markers Initiative (PPMI) cohorts. Linear regression multivariable models were applied to identify variables affecting Montreal Cognitive Assessment (MoCA) scores, accounting for age, sex, and years of education.

Clinical issues

Differences and contributors to global cognitive performance in the underrepresented Latinx Parkinson's disease population

Saar Anis, Henry Mauricio Chaparro-Solano, Thiago Peixoto Leal, Scott A. Sperling, Claire Sonneborn, Camila Callegari Piccinin, Miguel Inca-Martinez, Mario Cornejo-Olivas, Maryenela Illanes-Manrique, Pedro Chana-Cuevas, Paula Safie Awad, Ana Jimena Hernández-Medrano, Amin Cervantes-Arriaga, Artur F. S. Schuh, Carlos R. M. Rieder, Pedro Braga-Neto, Antonio Andrei da Silva Sena, Bruno Lopes Santos-Lobato, Emilia M. Gatto, Griselda J. Alvarado, Cesar L. Avila, Vitor Tumas, Maria Paula Foss, Vanderci Borges, Henrique Ballalai Ferraz, Jorge Luis Orozco Vélez, Beatriz Muñoz Ospina, Sonia Moreno, David Pineda, Julia Esther Rios Pinto, Patricio Olguín, Juan Cristobal Nuñez, Angel Viñuela, Alan O. Espinal-Martinez, Nicanor Mori, Koni Mejía-Rojas, Angel Medina-Colque, Ana Lucia Zuma Rosso, Celmir Vilaca, Edward Ochoa-Valle, Iván Cornejo-Herrera, Paula Reyes-Perez, Alejandra Lázaro-Figueroa, Anna Letícia de Moraes Alves, Rubens Gisbert Cury, Hubert H. Fernandez, Ignacio Mata & The Latin American Research consortium on the Genetics of Parkinson's Disease (LARGE-PD)

Abstract

Objective: Despite significant progress in understanding the factors influencing cognitive function in Parkinson's disease (PD), there is a notable gap in data representation for the Latinx population. This study aims to evaluate the contributors to and disparities in cognitive performance among Latinx patients with PD. **Methods:** A retrospective analysis was conducted based on cross-sectional data encompassing demographic, environmental, motor, and non-motor disease characteristics from the Latin American Research Consortium on the Genetics of PD (LARGE-PD) and the Parkinson's Progression Markers Initiative (PPMI) cohorts. Linear regression multivariable models were applied to identify variables affecting Montreal Cognitive Assessment (MoCA) scores, accounting for age, sex, and years of education. **Results:** The analysis comprised of 3,054 PD patients (2,041 from LARGE-PD and 1,013 from PPMI) and 1,303 Latinx controls. Latinx PD patients (mean age 63.0 $\pm$ 11.8, 56.8% male) exhibited a significantly lower average MoCA score ($p < .001$) compared to white non-Hispanic PD patients from PPMI (mean age 67.3 $\pm$ 9.9, 61.7% male). This difference persisted when comparing the Latinx PD to the Latinx controls (mean age 59.7 $\pm$ 9.3, 53.2% male; $p < .001$). Factors significantly associated with better MoCA scores in Latinx PD included: Unilateral symptom onset ($p < .009$) and higher educational attainment ($p < .001$). Conversely, those associated with worse scores included the use of dopamine agonists ($p = .01$), pramipexole use ($p = .01$), older age ($p < .001$), and a higher Hoehn and Yahr scale score ($p < .001$). **Conclusions:** Latinx PD patients demonstrated significantly lower cognitive scores compared to their white non-Hispanic PD counterparts and Latinx controls. These results highlight the importance of interpreting MoCA scores in a nuanced manner within diverse populations.

Keywords

Latinx, Montreal cognitive assessment, Parkinson's disease, cognitive function, cognitive dysfunction



(acceso libre)

Results

The analysis comprised of 3,054 PD patients (2,041 from LARGE-PD and 1,013 from PPMI) and 1,303 Latinx-controls. Latinx-PD patients (mean age 63.0 ± 11.8 , 56.8% male) exhibited a significantly lower average MoCA score ($p < .001$) compared to white Non-Hispanic PD patients from PPMI (mean age 67.5 ± 9.9 , 61.7% male). This difference persisted when comparing the Latinx-PD to the Latinx-controls (mean age 58.7 ± 9.3 , 33.2% male; $p < .001$). Factors significantly associated with better MoCA scores in Latinx-PD included unilateral symptom onset ($p = .009$), and higher educational attainment ($p < .001$). Conversely, those associated with worse scores included the use of dopamine agonists ($p = .01$), previous tobacco use ($p = .01$), older age ($p < .001$), and a higher Hoehn and Yahr scale score ($p < .001$).

Conclusions

Latinx-PD patients demonstrated significantly lower cognitive scores compared to their white non-Hispanic PD counterparts and Latinx-controls. These results highlight the importance of interpreting MoCA scores in a nuanced manner within diverse populations.

Research article

17. Differences in acute ischemic stroke treatment: A cross-sectional study from international Registry of Stroke Care Quality (RES-Q)

Mikulik R, Neto G, Sedani R, Ameriso SF, Mammadova N, Marchenko S, Martins S, Milanov I, Constanzo F, Muñoz M, Budincevic H, Šrámek M, Ramos C, Zakaria MF, Körv J, Tsivgoulis G, Szapary L, Pandian J, Nulhasanah A, Batayha W, Medukhanova S, Karbozova K, Miglane E, Vilionskis A, Kee HF, Gongora-Rivera F, Cantu Brito C, Groppa S, Ciobanu N, Paudel R, Abanto C, Collantes ME, San Jose MC, Kobayashi A, Gomes A, Tiu C, Shamalov N, Mijajlovic M, Gdovinová Z, Kroon L, Sohn SI, Moniche F, Towanabut S, Moskovko S, AlOmar A, Huy Thang N, Middleton S, Barrientos-Guerra JD / [View all authors and affiliations](#)

International Journal of Stroke, 2025

Abstract

Background

Stroke globally impacts mortality and disability. Compliance with international standards and evidence-based practices for acute stroke management would improve patient outcomes.

Objectives

We aimed to present a descriptive analysis of the quality of acute stroke care across different countries using the Registry of Stroke Care Quality (RES-Q).

Method

In a cross-sectional study, data from key quality indicators such as Emergency Medical Services (EMS) deployment rates, hospital arrival to imaging time (door-to-imaging: DIT), hospital arrival to thrombolysis time (door-to-needle: DNT), and Stroke Unit Care/Intensive Care Unit (SU/ICU) admission frequencies were examined. The analysis employed descriptive statistics and Spearman correlation tests.

Results

Of 334,184 patients from 1130 hospitals in 70 countries, 218,832 patients (65.5%) from 47 countries were diagnosed with acute ischemic stroke after exclusions. The number of patients per country ranged from 226 to 62,080. International variability in care quality was observed: EMS (7–97%); SU/ICU (12–100%); and median DIT (7–41 min); and DNT (20–75 min). IVT rates varied markedly across countries, ranging from <1% to 52%. Higher patient volumes were positively correlated with SU/ICU admission and negatively with DIT and DNT ($\rho = 0.10, -0.22, -0.42$, respectively).

Conclusion

This study demonstrates substantial international variation in the use of quality monitoring in clinical practice as well as in key indicators of acute ischemic stroke care, including intravenous thrombolysis rates and treatment timelines. The extent of variability highlights opportunities for benchmarking and targeted quality improvement efforts across diverse healthcare systems.



(suscripción o compra)

Disseminated cutaneous leishmaniasis with mucosal involvement in a patient with HTLV-1 infection: A case report

Highlights

- Rare HTLV-1/leishmaniasis co-infection; relapse post-Amphotericin B resolved with combo therapy.
- HTLV-1 co-infection complicates leishmaniasis management, requiring multidisciplinary approach and specialized follow-up.
- Combination therapy achieved complete remission in 30 days despite severe immunosuppression (disease and HTLV-1 co-infection).

Abstract

A case in Peru highlights the challenges of treating cutaneous leishmaniasis, a parasitic disease endemic to the region. A 44-year-old farmer presented with disseminated cutaneous leishmaniasis with mucosal involvement, further complicated by co-infection with HTLV-1, an unusual combination in Peru. Initial treatment with Amphotericin B was interrupted due to neuromuscular symptoms, causing disease recurrence. Serology later confirmed the HTLV-1 co-infection. After a comprehensive regimen including meglumine antimoniate, clindamycin, ceftriaxone, imiquimod, and pentoxifylline, the patient's lesions fully re-epithelialized within 30 days. The case emphasizes the necessity of treatment adherence and specialized follow-up to manage complex co-infections effectively.

Keywords

Cutaneous leishmaniasis, Macranthonia leishmaniasis, HTLV-1

Introduction

Leishmaniasis, caused by Leishmania parasites, is transmitted by infected hematophagous dipteran insects (Phlebotomus and Lutzomyia species). Leishmaniasis can affect the skin, mucous membranes, and viscera, with the specific form of the disease depending on the Leishmania species involved and the immune response of the host (1). Cutaneous

(suscripción o compra)

Case report

18. Disseminated cutaneous leishmaniasis with mucosal involvement in a patient with HTLV-1 infection

Sabina Mendivil-Tuchia de Tai^{a b}, Ursula Cáceres-Bernaola^{a b}, Silvy Sánchez Morales^{a b}, Isabel Farfán-Garcés^a, Wilfor Aguirre-Quispe^{c d}

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^d Instituto Nacional de Ciencias Neurológicas, Lima, Perú

Diagnostic Microbiology and Infectious Disease, February 2026

Abstract

A case in Peru highlights the challenges of treating cutaneous leishmaniasis, a parasitic disease endemic to the region. A 44-year-old farmer presented with disseminated cutaneous leishmaniasis with mucosal involvement, further complicated by co-infection with HTLV-1, an unusual combination in Peru. Initial treatment with Amphotericin B was interrupted due to neuromuscular symptoms, causing disease recurrence. Serology later confirmed the HTLV-1 co-infection. After a comprehensive regimen including meglumine antimoniate, clindamycin, ceftriaxone, imiquimod, and pentoxifylline, the patient's lesions fully re-epithelialized within 30 days. The case emphasizes the necessity of treatment adherence and specialized follow-up to manage complex co-infections effectively.

NEUROSURGERY **CASE REPORT**

Efficacy of Pressure Cooker Technique in Redo Embolization for High-Flow Torcular Dural Sinus Malformation

Frank Gleb Solis Chucos, MD, MPH¹, Rosa Lizbeth Ecos Quispe, MD, MPH^{2,3}, Mauro Toledo, MD¹, Melanie Walker, MD⁴, René Chapot, MD⁵

Introduction

Distal intra-arterial embolization (DIE) is a new, vascular anatomy characterized by various sizes and abnormal connections between dural sinuses and cerebral arteries, typically large, complex, and often involving the venous system. The diagnosis, treatment, and complications of this condition are still under investigation.

Case Report

A 44-year-old male patient presented with a large, high-flow torcular dural sinus malformation (tDSM) with high-flow fistulas, causing progressive venous hypertension and neurological symptoms. The patient had previously undergone a DIE procedure, but the malformation persisted. A redo embolization was performed using the pressure cooker technique (PCT) in a redo setting, allowing for high-pressure, targeted embolic delivery with minimized reflux, achieving near-complete occlusion and significant symptom relief. This case highlights PCT's potential to improve outcomes in multi-stage treatments of high-flow tDSM, reducing reflux and enhancing safety in technically demanding cases.

(acceso libre)

Case Report

19. Efficacy of Pressure Cooker Technique in Redo Embolization for High-Flow Torcular Dural Sinus Malformation

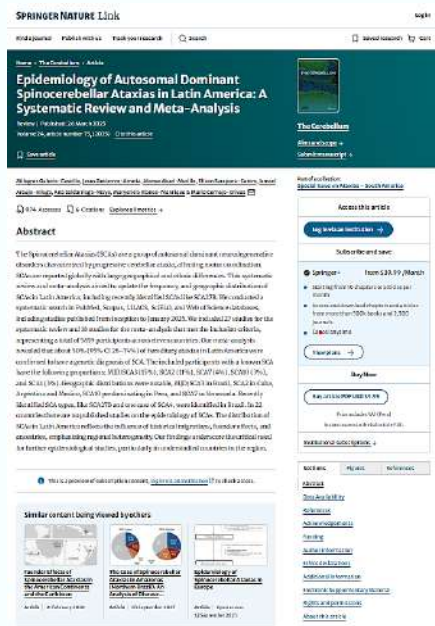
Frank Gleb Solis Chucos, MD, MPH¹, Rosa Lizbeth Ecos Quispe, MD, MPH^{2,3}, Mauro Toledo, MD¹, Melanie Walker, MD⁴, René Chapot, MD⁵

¹Department of Endovascular Neurosurgery, Instituto Nacional de Salud del Niño-San Borja, Lima, Perú ²Department of Vascular Neurology, Instituto Nacional de Ciencias Neurológicas, Lima, Perú ³Department of Medicine, Universidad Nacional Federico Villarreal, Lima, Perú ⁴Department of Neurological Surgery, University of Washington, Seattle, WA, USA ⁵Department of Interventional Neuroradiology, Alfred Krupp Hospital Rüttenscheid, Essen, Germany

Neurointervention, 2025 Mar;20(1):32-36

Abstract

Torcular dural sinus malformations (tDSMs) with high-flow fistulas pose complex management challenges due to their vascularity and the delicate neuroanatomy involved. This report presents the case of a child with tDSM and hydrocephalus, who underwent 3 staged embolization procedures but required a redo intervention due to residual malformation and venous hypertension. Utilizing the pressure cooker technique (PCT) in a redo setting allowed for high-pressure, targeted embolic delivery with minimized reflux, achieving near-complete occlusion and significant symptom relief. This case highlights PCT's potential to improve outcomes in multi-stage treatments of high-flow tDSM, reducing reflux and enhancing safety in technically demanding cases.



(suscripción o compra)

Article

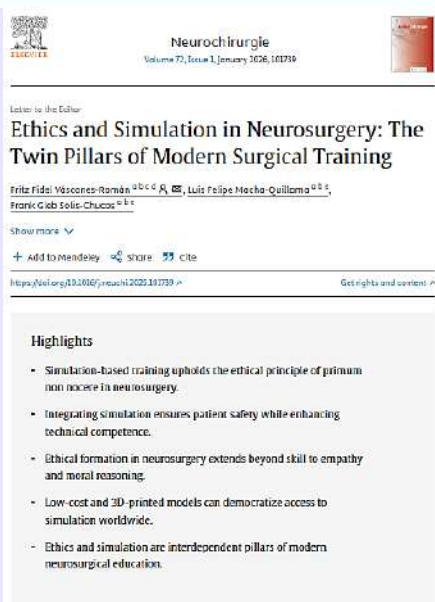
20. Epidemiology of Autosomal Dominant Spinocerebellar Ataxias in Latin America: A Systematic Review and Meta-Analysis

Milagros Galecio-Castillo, Jesus Gutierrez-Arratia, Alonso Abad-Murillo, Elison Sarapura-Castro, Ismael Araujo-Aliaga, Ana Saldarriaga-Mayo, Maryenela Illanes-Manrique & Mario Cornejo-Olivas

Cerebellum 24,75, 2025

Abstract

The Spinocerebellar Ataxias (SCAs) are a group of autosomal dominant neurodegenerative disorders characterized by progressive cerebellar ataxia, affecting motor coordination. SCAs are reported globally with large geographical and ethnic differences. This systematic review and meta-analysis aimed to update the frequency, and geographic distribution of SCAs in Latin America, including recently identified SCAs like SCA27B. We conducted a systematic search in PubMed, Scopus, LILACS, SciELO, and Web of Science databases, including studies published from inception to January 2025. We included 27 studies for the systematic review and 18 studies for the meta-analysis that met the inclusion criteria, representing a total of 5859 participants across eleven countries. Our meta-analysis revealed that about 50% (95% CI 26–74%) of hereditary ataxias in Latin America were confirmed to have a genetic diagnosis of SCA. The included participants with a known SCA have the following proportions: MJD/SCA3 (15%), SCA2 (11%), SCA7 (4%), SCA10 (3%), and SCA1 (3%). Geographic distributions were notable, MJD/SCA3 in Brazil, SCA2 in Cuba, Argentina and Mexico, SCA10 predominating in Peru, and SCA7 in Venezuela. Recently identified SCA types, like SCA27B and one case of SCA4, were identified in Brazil. In 22 countries there are no published studies on the epidemiology of SCAs. The distribution of SCAs in Latin America reflects the influence of historical migrations, founder effects, and ancestries, emphasizing regional heterogeneity. Our findings underscore the critical need for further epidemiological studies, particularly in understudied countries in the region.



(suscripción o compra)

Letter to the editor

21. Ethics and Simulation in Neurosurgery: The Twin Pillars of Modern Surgical Training

Fritz Fidel Váscones-Román a b c d, Luis Felipe Macha-Quillama a b e, Frank Gleb Solis-Chucos a b e

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b Universidad Peruana Cayetano Heredia, Lima, Peru
c Astrocyte, Neurosurgical Research Group, Boston, MA, United States of America
d Váscones's Lab, Lima, Peru
e Clínica Internacional, Lima, Peru

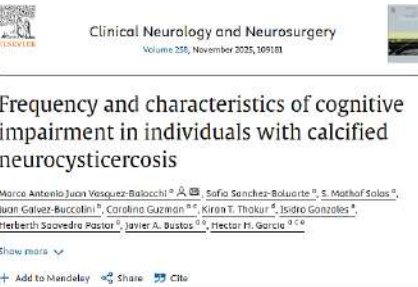
Neurochirurgie, Volume 72, enero 2026

Section snippets**Simulation as an ethical and pedagogical tool**

Simulation-based training allows neurosurgeons in formation to acquire and refine operative skills within a safe, controlled environment. Studies consistently show that residents trained with simulators demonstrate superior technical performance compared to those relying solely on operating-room exposure [4,5]. Multiple modalities exist—from synthetic and cadaveric models to 3D-printed replicas and immersive virtual reality (VR) simulators. While each has unique advantages, recent technological

Ethical formation beyond technique

Beyond technical mastery, simulation promotes moral reasoning, professionalism, and reflective practice. Simulation-based curricula can integrate ethical decision-making, communication, and crisis management, allowing residents to rehearse scenarios such as intraoperative complications, disclosure of adverse events, or informed consent discussions [10]. Structured debriefings—where participants analyze both technical and ethical choices—strengthen moral awareness and foster a culture of...

	Challenges and barriers Despite its benefits, simulation faces barriers to global adoption. High-fidelity simulators and VR systems remain expensive, particularly for resource-limited institutions. A lack of standardized curricula, validated assessment tools, and trained faculty further hinders implementation [4,6]. Cultural inertia also persists; in some academic cultures, simulation is still considered secondary to “real” operative experience. Overcoming this requires faculty engagement and institutional support
 Expanded ATTCT-ATXN10 alleles in healthy Peruvian population I. Araujo-Aliaga^{1,2,3}, K. Milla-Neyra², C. Manrique-Enciso², E. Sarapura-Castro^{1,2}, M. Illanes-Manrique^{2,3}, D. Veliz-Otani^{2,4}, A. Saldarriaga-Mayo^{2,3}, A. Medina-Colque⁵, J. Rios-Pinto⁶, I. Cornejo-Herrera⁷, A. Rivera-Valdivia^{2,3}, I. Mata⁸, D. Loesch⁹, L. Lozano-Vasquez^{2,9}, T. Bordia¹⁰, T. O'Connor⁴, B. Schüle¹⁰, M. Cornejo-Olivera^{1,2,3} References (0) Cited by (0) (suspcripción o compra)	Journal article 22. Expanded ATTCT-ATXN10 alleles in healthy peruvian population I. Araujo-Aliaga^{1,2,3}, K. Milla-Neyra², C. Manrique-Enciso², E. Sarapura-Castro^{2,3}, M. Illanes-Manrique^{2,3}, D. Veliz-Otani^{2,4}, A. Saldarriaga-Mayo^{2,3}, A. Medina-Colque⁵, J. Rios-Pinto⁶, I. Cornejo-Herrera⁷, A. Rivera-Valdivia^{2,3}, I. Mata⁸, D. Loesch⁴, L. Lozano-Vasquez^{2,9}, T. Bordia¹⁰, T. O'Connor⁴, B. Schüle¹⁰, M. Cornejo-Olivera^{1,2,3} ¹Neurogenetics Research Unit, Fundacion San Marcos para el Desarrollo de la Ciencia y la Cultura, Lima, Peru ²Neurogenetics Research Center, Instituto Nacional de Ciencias Neurológicas, Lima, Peru ³Neurogenetics Working Group, Universidad Científica del Sur, Lima, Peru ⁴Institute for Genome Sciences, School of Medicine, University of Maryland, Baltimore, Maryland, USA ⁵Dirección Regional de Salud de Puno, Puno, Peru ⁶Universidad Peruana Los Andes, Huancayo, Peru ⁷Universidad Privada de Tacna, Tacna, Peru ⁸Genomic Medicine Institute, Lerner Research Institute, Cleveland Clinic, Cleveland, Ohio, USA ⁹Universidad Peruana de Ciencias Aplicadas, Lima, Peru ¹⁰Department of Pathology, Stanford University School of Medicine, California, USA Parkinsonism & Related Disorders, Vol. 134, 16 April 2025 Antecedentes La ataxia espinocerebelosa tipo 10 (SCA10) es un trastorno neurodegenerativo que afecta predominantemente a latinoamericanos con ascendencia amerindia, causado por una expansión de repeticiones ATTCT en el gen ATXN10. Los individuos sanos tienen entre 9 y 32 repeticiones ATTCT, mientras que los individuos con SCA10 presentan 280 o más repeticiones ATTCT. Sin embargo, hallazgos recientes muestran alelos expandidos (>32 repeticiones) en amerindios peruanos sanos, con significado incierto. Además, el alelo G en el SNP rs41524547 se considera un factor de riesgo para la SCA10. Este estudio tiene como objetivo genotipificar el microsatélite ATTCT-ATXN10 y explorar la frecuencia del SNP rs41524547 en las subpoblaciones mestizas y amerindias peruanas.
 Frequency and characteristics of cognitive impairment in individuals with calcified neurocysticercosis Marco Antonio Juan Vasquez-Baiocchi^a, Sofia Sanchez-Boluarte^a, S. Mathof Salas^a, Juan Galvez-Buccolini^b, Carolina Guzman^{a,c}, Kiran T. Thakur^d, Isidro Gonzales^e, Herberth Saavedra Pastor^e, Javier A. Bustos^{a,e}, Hector H. Garcia^{a,c,e} Highlights • Memory-related issues are a common, understudied complaint in neurocysticercosis. • Cognitive impairment was more prevalent in calcified neurocysticercosis. • Calcifications in the left temporal lobe are associated with lower cognitive scores. (suspcripción o compra)	Research article 23. Frequency and characteristics of cognitive impairment in individuals with calcified neurocysticercosis Marco Antonio Juan Vasquez-Baiocchi^a, Sofia Sanchez-Boluarte^a, S. Mathof Salas^a, Juan Galvez-Buccolini^b, Carolina Guzman^{a,c}, <u>Kiran T. Thakur^d</u>, Isidro Gonzales^e, Herberth Saavedra Pastor^e, <u>Javier A. Bustos^{a,e}</u>, <u>Hector H. Garcia^{a,c,e}</u> ^aCenter for Global Health, Universidad Peruana Cayetano Heredia, Avenida Honorio Delgado 430, Lima, Peru ^bDepartment of Psychiatry, Clinica Angloamericana, Calle Alfredo Salazar 350, Lima, Peru ^cDepartment of International Health, John Hopkins Bloomberg School of Public Health, 615 N. Wolfe Street, Baltimore, MD, United States ^dProgram in Neuroinfectious Diseases, Department of Neurology, Columbia University Irving Medical Center, New York Presbyterian Hospital, 630 W, 168th Street, New York, NY, United States ^eCentro Básico de Investigación (CBI) en Exámenes Auxiliares en Parasitosis del Sistema Nervioso (Laboratorio de Cisticercosis), Instituto Nacional de Ciencias Neurológicas, Jirón Ancash, Lima 1271, Peru Clinical Neurology and Neurosurgery, 2025 Abstract Objective Neurocysticercosis (NCC) is the most common helminthic infection of the central nervous system (CNS). When NCC calcifies, it causes symptoms including memory impairment. Here we will assess the frequency and characteristics of cognitive impairment using screening tools in patients with calcified NCC.

Methods

We performed a cross-sectional study in participants of an ongoing cohort of patients with calcified NCC, who were administered the Mini-Mental State Examination (MMSE) and Memory Alteration Test (MAT) simultaneously. We analyzed these test scores in relation to sociodemographic, clinical, and neuroimaging characteristics.

Results

We included 336 individuals, with an average age of 43.6 ± 14.8 years, 56 % of whom were female. The median scores for MMSE and MAT were 27 (IQR 25-28) and 41 (IQR 36-45), respectively. We observed cognitive impairment in 43 (12.8 %) participants with MMSE and 90 (26.8 %) with MAT. Among the 57 participants aged 60 years and older, 17 (29.8 %) had cognitive impairment on MMSE and 29 (50.9 %) on MAT. Lower scores were associated with older age ($p < 0.001$ for both tests), and the presence of left temporal lobe calcifications ($p < 0.001$ for both), associations that persisted after multivariate adjustment ($p = 0.017$ for MMSE and $p = 0.043$ for MAT).

Conclusion

In this cohort, we found that low cognitive scores are frequently observed in patients with calcified NCC (12.8 % by MMSE, 26.8 % by MAT) and are associated with older age and calcifications in the left temporal lobe. Further studies with appropriate control groups are needed to assess these associations beyond screening tools and examine other contributing factors.

Poster presentation

24. Generalizability of Alzheimer's disease plasma biomarkers phospho-Tau and beta amyloid in individuals of diverse genetic ancestries

Anthony J Griswold^{1,2}, Farid Rajabli^{2,3}, Tianjie Gu¹, Jamie Arvizu¹, Charles G Golightly¹, Patrice L Whitehead¹, Kara L Hamilton-Nelson³, Larry D Adams³, Katrina Celis³, Jose Javier Sanchez³, Pedro R Mena³, Takiyah D Starks⁴, Maryenela Z Illanes-Manrique⁵, Concepcion Silva-Vergara⁶, Michael L Cuccaro^{2,3}, Jeffery M Vance^{2,3}, Briseida E Feliciano-Astacio⁶, Mario Comejo-Olivas⁵, Goldie S Byrd⁴, Gary Beecham^{2,3}, Jonathan L Haines⁷, Margaret A Pericak-Vance^{2,8}

¹John P.Hussman Institute for Human, Genomics,University of Miami Miller School of Medicine, Miami,FL,USA,Miami,FL,USA

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Alzheimer's & Dementia, 2025 Jan 9;20(Suppl 2)

Abstract

Background

Plasma concentrations of phosphorylated threonine-181 of Tau (pTau181) and the ratio of amyloid beta isoforms Aβ42/Aβ40 are biomarkers for differential diagnosis and preclinical detection of Alzheimer disease (AD). However, assessment of the utility of these biomarkers has been in non-Hispanic, European individuals. Given differences in AD risk across populations, generalizability of these findings is not assured in individuals of diverse ancestries. Here we evaluate the levels of plasma pTau181 and Aβ42/Aβ40 and assess their utility in discriminating clinically diagnosed AD from cognitively unimpaired (CU), age-matched controls in ancestrally diverse, admixed cohorts.

Method

We measured pTau181 and Aβ42/Aβ40 with Simoa chemistry using the pTau181 AdvantageV2 and NEUROLOGY 3-PLEX A assays, respectively. Our cohorts consisted of: 923 African Americans (319 AD, 604 CU), 149 Peruvians (49 AD, 100 CU), and 667 Caribbean Hispanics (613 Puerto Ricans (288 AD,

DOI: 10.1093/alzdis/alz001

Alzheimer's & Dementia
The Journal of the Alzheimer's Society

BIOMARKERS
POSTER PRESENTATION

BIOMARKERS (NON-NEUROIMAGING)

Generalizability of Alzheimer's disease plasma biomarkers phospho-Tau and beta amyloid in individuals of diverse genetic ancestries

Anthony J. Griswold^{1,2}, Farid Rajabli^{2,3}, Tianjie Gu¹, Jamie Arvizu¹, Charles G. Golightly¹, Patrice L. Whitehead¹, Kara L. Hamilton-Nelson³, Larry D. Adams³, Katrina Celis³, Jose Javier Sanchez³, Pedro R. Mena³, Takiyah D. Starks⁴, Maryenela Z. Illanes-Manrique⁵, Concepcion Silva-Vergara⁶, Michael L. Cuccaro^{2,3}, Jeffery M. Vance^{2,3}, Briseida E. Feliciano-Astacio⁶, Mario Comejo-Olivas⁵, Goldie S. Byrd⁴, Gary Beecham^{2,3}, Jonathan L. Haines⁷, Margaret A. Pericak-Vance^{2,8}

¹John P.Hussman Institute for Human, Genomics,University of Miami Miller School of Medicine, Miami,FL,USA,Miami,FL,USA
²Dr. John T. Mac Donald Foundation, Department of Human Genetics,University of Miami Miller School of Medicine, Miami,FL, USA
³John P.Hussman Institute for Human, Genomics,University of Miami Miller School of Medicine, Miami,FL,USA
⁴Maya Angelou Center for Health Equity, Wake Forest University School of Medicine, Winston-Salem,NC,USA
⁵Neurogenetics Research Center, Instituto Nacional de Ciencias Neurológicas,,Lima, Peru
⁶Universidad Central del Caribe,Bayamón,PR, USA
⁷Department of Population and Quantitative, Health Sciences, Institute for Computational Biology, Case Western Reserve University, Cleveland, OH,USA 81501 NW 10th Avenue, Miami, FL, USA

Abstract
Background: Plasma concentrations of phosphorylated threonine-181 of Tau (pTau181) and the ratio of amyloid beta isoforms Aβ42/Aβ40 are biomarkers for differential diagnosis and preclinical detection of Alzheimer disease (AD). However, assessment of the utility of these biomarkers has been in non-Hispanic, European individuals. Given differences in AD risk across populations, generalizability of these findings is not assured in individuals of diverse ancestries. Here we evaluate the levels of plasma pTau181 and Aβ42/Aβ40 and assess their utility in discriminating clinically diagnosed AD from cognitively unimpaired (CU), age-matched controls in ancestrally diverse, admixed cohorts.
Method: We measured pTau181 and Aβ42/Aβ40 with Simoa chemistry using the pTau181 AdvantageV2 and NEUROLOGY 3-PLEX A assays, respectively. Our cohorts consisted of: 923 African Americans (319 AD, 604 CU), 149 Peruvians (49 AD, 100 CU), and 667 Caribbean Hispanics (613 Puerto Ricans (288 AD, 329 CU), 289 CU). Linear mixed-effects regression models adjusted for age, sex, population substructure and admixture. Biomarkers and receptors were tested for their utility in discriminating AD from cognitively unimpaired (CU), age-matched controls in ancestrally diverse, admixed cohorts.

This work was supported by the National Institutes of Health (NIH) under the terms of the Cooperative Agreement (P50 AG056000) and the Alzheimer's Disease Research Center (ADRC) at the University of Miami. The authors thank the participants and staff of the Alzheimer's Disease Research Center (ADRC) at the University of Miami for their contributions to this study.

Alzheimer's & Dementia, 2025 Jan 9;20(Suppl 2)



(acceso libre)

325 CU) and 54 Cubans (24 AD, 30 CI)). Linear mixed-effect regression models adjusted for age, sex, population substructure and relatedness followed by Bonferroni correction was applied to identify biomarker differences. Diagnostic performance and receiver operator characteristic (ROC) curves were created from logistic regression models.

Result

Plasma pTau181 concentrations did not differ across any of the cohorts within AD or CU. There was a clear elevation of pTau181 concentration in AD compared to CU ($p < 1.7 \times 10^{-13}$) taking into account all individuals and in each cohort separately (African Americans, $p = 1.9 \times 10^{-8}$; Caribbean Hispanics, $p = 3.1 \times 10^{-7}$; Peruvians, $p = 0.05$). There was no significant difference in the plasma A β 42/A β 40 ratio, however there was a trend towards a decreasing ratio in AD. Using the area under the ROC, pTau181 was more accurate at predicting status than the A β 42/A β 40 ratio, but the classification improved when both biomarkers were combined.

Conclusion

These results suggest AD biomarkers are generalizable across ancestries, with baseline values being consistent across diverse populations. Ultimately, combining genomic, biomarker, and social and environmental data from diverse individuals will increase understanding of genetic risk and refine clinical diagnoses in individuals of diverse ancestries.

Research article

25. Generalizability of tau and amyloid plasma biomarkers in Alzheimer's disease cohorts of diverse genetic ancestries.

Anthony J. Griswold^{1,2}; Farid Rajabli^{1,2}; Tianjie Gu¹; Jamie Arvizu¹; Charles G. Golightly¹; Patrice L. Whitehead¹; Kara L. Hamilton-Nelson¹; Larry D. Adams¹; Jose J. Sanchez¹; Pedro R. Mena¹; Takiyah D. Starks³; Maryenelalllanes-Manrique⁴; Concepcion Silva⁵; William S. Bush^{6,7}; Michael L. Cuccaro^{1,2}; Jeffery M. Vance^{1,2}; Mario R. Cornejo-Olivas⁴; Briseida E. Feliciano-Astacio⁵; Goldie S. Byrd³; Gary W. Beecham^{1,2}; Jonathan L. Haines^{6,7}; Margaret A. Pericak-Vance^{1,2}

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Alzheimer's & Dementia, 2025 Mar;21(3)

Abstract

Introduction

Plasma phosphorylated threonine-181 of Tau and amyloid beta are biomarkers for differential diagnosis and preclinical detection of Alzheimer disease (AD). Given differences in AD risk across diverse populations, generalizability of existing biomarker data is not assured.

Methods

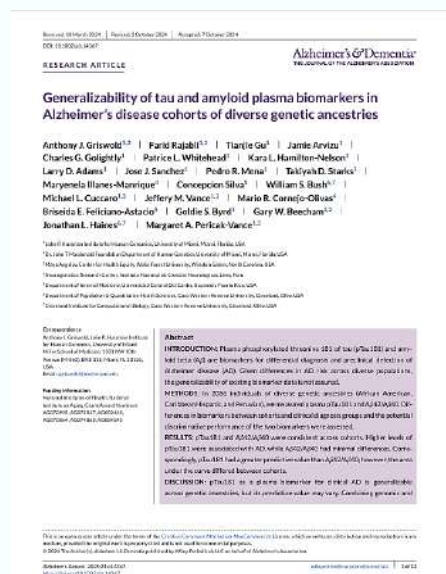
In 2,086 individuals of diverse genetic ancestries (African American, Caribbean Hispanic, and Peruvians) we measured plasma pTau-181 and A β 42/A β 40. Differences in biomarkers between cohorts and clinical diagnosis groups and the potential discriminative performance of the two biomarkers were assessed.

Results

pTau-181 and A β 42/A β 40 were consistent across cohorts. Higher levels of pTau181 were associated with AD while A β 42/A β 40 had minimal differences. Correspondingly, pTau-181 had greater predictive value than A β 42/A β 40, however, the area under the curve differed between cohorts.

Discussion

pTau-181 as a plasma biomarker for clinical AD is generalizable across genetic ancestries, but predictive value may differ. Combining genomic and biomarker data from diverse individuals will increase understanding of genetic risk and refine clinical diagnoses.



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Preprint

26. Genotype-phenotype association study conducted on LARGE-PD reveals novel loci associated with Parkinson's Disease

Thiago P Leal, Emily Waldo, Felipe Duarte-Zambrano, Miguel Inca-Martinez, Janvi Ramchandra, Henry Mauricio Chaparro-Solano, Anna E Anello, Victor Borda, Mateus Henrique Gouveia, Daniel Teixeira-dos-Santos, Paula Reyes-Pérez, Emilia Mabel Gatto, View ORCID Profile Bruno Lopes Santos-Lobato, Gracivane Eufaseo, Grace Helena Letro, Gonzalo Arboleda, Oscar Bernal-Pacheco, Jorge L Orozco, Beatriz Munoz, Pedro Chana-Cuevas, David Aguilon, Sonia Moreno, Gabriel Torrealba-Acosta, Tanya Lobo-Prada, Valentina Muller, C Matias Lopez Razquin, Pedro Braga-Neto, Reyna M Durón, View ORCID Profile Mayela Rodríguez-Violante, Ana Jimena Hernández-Medrano, Amin Cervantes-Arriaga, Daniel Martínez-Ramírez, View ORCID Profile Artur F S Schuh, View ORCID Profile Carlos Roberto de Mello Rieder, Mario Cornejo-Olivas, Julia Rios-Pinto, Angel C Medina, Ivan Cornejo-Herrera, Koni Mejia-Rojas, Angel Vinuela, Vitor Tumas, Angela Vieira Pimentel, Vanderi Borges, Cesar L Avila, Patricio Olguin, Alicia Colombo, Juan Cristobal Nuñez, View ORCID Profile Alejandra Medina-Rivera, View ORCID Profile Alejandra E. Ruiz-Contreras, View ORCID Profile Saraél Alcauter, Elena Dieguez, Karen Nuytemans, Global Parkinson's Genetics Program, View ORCID Profile Ignacio F Mata the Latin American Research Consortium on the Genetics of Parkinson's Disease (LARGE-PD)

medRxiv, 2025

Abstract**Background**

The Latin American Research Consortium on the Genetics of Parkinson's Disease (LARGE-PD) is a multicenter collaboration aimed at understanding the genetic architecture of Parkinson's disease (PD) in this underrepresented population using data from 15 countries across the Americas and the Caribbean. In this study, we conducted the largest genome-wide association studies (GWAS) for PD susceptibility in Latin Americans.

Methods

We analyzed genotype data from LARGE-PD Phase 1 (n = 1,498) and Phase 2 (n = 4,401) using multiple GWAS approaches: SAIGE, which incorporates a genetic relationship matrix in the model; ATT, which includes global ancestry on the model; TRACTOR, which splits allele dosages by ancestry to detect ancestry-specific risk loci; and admixture mapping. We also assessed linkage disequilibrium (LD) patterns and performed Meta-Regression of Multi-Ancestry Genetic Association (MR-MEGA), integrating data from both LARGE-PD phases and two South Asian GWAS.

Results

We identified PD-associated loci on chromosomes 1 and 4. Our results replicated previous findings, including the well-established SNCA variant rs356182-A (OR = 1.517, p = 1.62×10⁻¹⁶). Notably, we identified a locus in ITPKB (rs117185933-A, OR = 1.75, p = 3.8×10⁻¹²), which had the highest CADD Phred score (17.92, top ~3% most deleterious) among all candidate variants, suggesting strong functional relevance. Functional annotation predicted that this variant may create a premature start codon in the 5' UTR of ITPKB. Although rs117185933-A is in high LD (r² > 0.8) with a variant previously reported by Kishore et al., our LD analysis and MR-MEGA results indicate that this signal is correlated with ancestry heterogeneity and likely represents an independent PD risk locus and a novel putative causal variant. This variant is most frequent in Peruvians from the 1000 Genomes Project (MAF = 0.20) and more common in admixed American populations in gnomAD (MAF = 0.0835), but nearly absent in non-Finnish Europeans (MAF = 0.0002).

Conclusion

We identified PD-associated variants in SNCA and ITPKB, the latter not previously reported in European-ancestry studies. The ITPKB variant may lead to a start codon gain in a gene with known protective effects against α-synuclein aggregation in vivo and in vitro models. These findings underscore the critical importance of including underrepresented populations in genetic research to uncover ancestry-specific risk loci and advance precision medicine for Parkinson's disease.

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Genotype-phenotype association study conducted on LARGE-PD reveals novel loci associated with Parkinson's Disease.

Thiago P Leal¹, Emily Waldo¹, Felipe Duarte-Zambrano^{1,2}, Miguel Inca-Martinez¹, Janvi Ramchandra¹, Henry Mauricio Chaparro-Solano¹, Anna E Anello¹, Victor Borda^{1,3}, Mateus Henrique Gouveia¹, Daniel Teixeira dos Santos¹, Paula Reyes-Pérez¹, Emilia Mabel Gatto¹, Bruno Lopes Santos-Lobato^{1,4}, Gracivane Eufaseo¹, Grace Helena Letro^{1,5}, Gonzalo Arboleda^{1,6}, Oscar Bernal Pacheco^{1,7,8}, Jorge L Orozco^{1,9}, Beatriz Munoz^{1,10}, Pedro Chana-Cuevas^{1,11}, David Aguilon^{1,12}, Sonia Moreno^{1,13}, Gabriel Torrealba-Acosta^{1,14}, Tanya Lobo-Prada^{1,15}, Valentina Muller^{1,16}, C Matias Lopez Razquin^{1,17}, Pedro Braga-Neto^{1,18}, Reyna M Durón^{1,19}, Mayela Rodríguez-Violante^{1,20}, Ana Jimena Hernández-Medrano^{1,21}, Amin Cervantes-Arriaga^{1,22}, Daniel Martínez-Ramírez^{1,23}, Artur F S Schuh^{1,24}, Carlos Roberto de Mello Rieder^{1,25}, Mario Cornejo-Olivas^{1,26}, Julia Rios-Pinto^{1,27}, Angel C Medina^{1,28}, Ivan Cornejo-Herrera^{1,29}, Koni Mejia-Rojas^{1,30}, Angel Vinuela^{1,31}, Vitor Tumas^{1,32}, Angela Vieira Pimentel^{1,33}, Vanderi Borges^{1,34}, Cesar L Avila^{1,35}, Patricio Olguin^{1,36}, Alicia Colombo^{1,37}, Juan Cristobal Nuñez^{1,38}, Alejandra Medina-Rivera^{1,39}, Alejandra E Ruiz-Contreras^{1,40}, Saraél Alcauter^{1,41}, Elena Dieguez^{1,42}, Karen Nuytemans^{1,43}, Global Parkinson's Genetics Program, Ignacio F Mata^{1,44} on behalf of the Latin American Research Consortium on the Genetics of Parkinson's Disease (LARGE-PD).

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(suscripción o compra)

Rapid communication

27. In vitro system for Taenia solium cysts culture and growth: progress toward a neurocysticercosis model

Miguel A. Orrego, Oscar H. Del Brutto, Carlos M. Vasquez, Theodore E. Nash, Hector H. Garcia &for The Cysticercosis Working Group in Peru

Future Microbiology, Volume 20, 2025

Abstract

Aims

Subarachnoid neurocysticercosis is an aggressive form of Taeniasoliuminfection characterized by uncontrolled expansion of subarachnoid cysts. This study aims to investigate the role of exogenous lipidsand TGF-βin the proliferation of germinative cells and bladder wall expansion, providing insights into the biological mechanisms underlying cyst growth.

Material & Methods

Germinative cells and bladder wall sections discarded after surgical resection of subarachnoid cysts in humans were cultured in vitro under axenic conditions. The effects of TGF-βand exogenous lipids, both individually and combined, were assessed through proliferation assays and morphological analysis to determine their role in cyst expansion.

Results

TGF-β-mediatedsignaling was found to be crucial for germinative cell replication, with lipid supply further enhancing bladder wall expansion. Long-term viability of cultured bladder wall sections was successfully maintained, enabling extended analysis of cyst biology.

Conclusions

Thisinvitrosystem provides a powerful platform for characterizing the biology of subarachnoid cysts and for testing new therapeutic agents aimed at improving the treatment of human neurocysticercosis.



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Artículo de investigación

28. Incidencia de Pacientes con Síndrome de Guillain Barré en el Servicio de Neurointensivo del Instituto Nacional de Ciencias Neurológicas / Incidence of Patients with Guillain-Barré Syndrome in the Neurointensive Care Unit of the National Institute of Neurological Sciences

Néstor Ríos Vigil1

1Médico Neurólogo del Servicio de Neurointensivo del Instituto Nacional de Ciencias Neurológicas, Lima, Perú

SAGA Revista Científica Multidisciplinar 2(2):194-202, April 2025.

Resumen

Se trata de 4 casos del Síndrome de Guillain Barré, 2 del Tipo AMAN (Variante axonal motora aguda), 1 de tipo ASMAN (Variante axonal sensitivo-motora aguda) y 1 de tipo AIDP (variante inflamatoria aguda desmielinizante), durante el año 2024, en el Servicio de Neurointensivo del Instituto Nacional de Ciencias Neurológicas (INCN), la mayoría con un tiempo de enfermedad de 4-5 días, los 4 de sexo masculino, con uso de ventilación mecánica y de inmunoglobulina, sin compromiso de pares craneales, con complicaciones respiratorias, abdominales, renales, de medio interno, psiquiátricas, disautonomía y otras. También hubo un pródromo respiratorio o abdominal, el cual fue visto por la oficina de epidemiología, la evolución fue favorable con la interconsulta a medicina física y rehabilitación.

Article

29. Machine Learning approaches for predicting Levodopa-Induced Dyskinesias in Latino Populations

H.M. Chaparro-Solano ^{1 2}, M. Inca-Martínez ², G. Alvarado ³, S. Alcauter ⁴, G. Arboleda ⁵, C.L. Avila ³, P. Chana-Cuevas ⁶, M. Cornejo-Olivas ^{7 8}, E. Fernandez ⁹, E. Gatto ¹⁰, M. Jimenez-Del-Rio ¹¹, D. Aguillon ¹¹, M. Kauffman ¹², D. Martinez-Ramirez ¹³, V. Muller ¹⁴, P. Olguin ^{15 16}, J.L. Orozco-



Parkinsonism & Related Disorders
Volume 134, May 2025, 107549

Machine Learning approaches for predicting Levodopa-Induced Dyskinesias in Latino Populations

H.M. Chaparro-Solano^{1,2}, V. Irujo-Martínez³, G. Alvarado⁴, S. Alcazar⁵, G. Arboleda⁶, C. Alvilé⁷, P. Chana-Cuevas⁸, M. González-Olivas⁹, E. Fernández¹⁰, E. Gatto¹¹, M. Jiménez-Del-Río¹², D. Aguilón¹³, M. Kaufman¹⁴, G. Martínez-González¹⁵, V. Müller¹⁶, A. Ojeda¹⁷, J. L. Ojeda-Velasco¹⁸, S. Peña¹⁹, M. Rodríguez-Velante²⁰, A. Vimala²¹, J. S. Mata^{1,2}

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(suscripción o compra)

Velez^{17,18}, S. Peña¹⁹, M. Rodríguez-Velante²⁰, A. Vinuela²¹, I.F. Mata^{1,2}, The Latin American Research consortium on the Genetics of Parkinson's Disease (LARGE-PD)

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Parkinsonism & Related Disorders, Volume 134, 2025

Antecedentes

La discinesia inducida por levodopa (DIL) sigue siendo una de las necesidades no cubiertas más importantes en la enfermedad de Parkinson (EP), ya que afecta la calidad de vida (CdV) de las personas con EP y aumenta el costo del tratamiento. A pesar de estar asociada con el medicamento de primera línea para la EP y su relativa alta frecuencia, actualmente no existe ninguna herramienta disponible para predecir la aparición de DIL. Este estudio tiene como objetivo desarrollar un modelo de aprendizaje automático (AA) utilizando datos clínicos y demográficos del consorcio latinoamericano de investigación sobre la genética de la EP (LARGE-PD) para predecir la aparición de DIL.



Neurology Journals, Issue 105 (7), 2025

Mediation Analysis to Identify Individual Component Effects of the Care Bundle to Improve Functional Outcome After ICH in the INTERACT3 Study

Xinwen Ren, Qiang Li, Menglu Ouyang, Laurent Billot, Xiaoying Chen, Paula Munoz Venturelli, Asita de Silva, Thang Huy Nguyen, Kolawole W. Wahab, Jeyaraj Durai Pandian, Mohammad Wasay, Octavio M. Pontes-Neto, Carlos Abanto, Antonio Arauz, Chao You, Lu Ma, Xin Hu, Craig Anderson, Xia Wang, and Lili Song as the INTERACT3 Investigators

[Authors Info & Affiliations](#)

Abstract

Background and Objectives

The third Intensive Care Bundle with Blood Pressure Reduction in Acute Cerebral Hemorrhage Trial (INTERACT3) showed that the implementation of a care bundle improves the functional outcome of patients who experience acute intracerebral hemorrhage (ICH). However, uncertainty exists over the relative contribution of each component of the care bundle to the benefit.

Methods

INTERACT3 used a stepped-wedge, cluster randomized controlled trial design, which was conducted in 5 lower and 4 upper middle-income countries and 1 high-income country. Compared with usual care, there was a statistically significant beneficial effect of the intervention, a care bundle comprising early control of elevated blood pressure (BP), glucose, and temperature and reversal of warfarin-related anticoagulation, on the primary outcome of functional recovery of ICH. We performed a model-based causal mediation analysis to assess the contribution of each component of the care bundle to the overall effect, as measured by the modified

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Research article

30. Mediation Analysis to Identify Individual Component Effects of the Care Bundle to Improve Functional Outcome After ICH in the INTERACT3 Study
Xinwen Ren, Qiang Li, Menglu Ouyang, Laurent Billot, Xiaoying Chen, Paula Munoz Venturelli, Asita de Silva, Thang Huy Nguyen, Kolawole W. Wahab, Jeyaraj Durai Pandian, Mohammad Wasay, Octavio M. Pontes-Neto, Carlos Abanto, Antonio Arauz, Chao You, Lu Ma, Xin Hu, Craig Anderson, Xia Wang, and Lili Song as the INTERACT3 Investigators
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Neurology Journals, Issue 105 (7), 2025

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	Results A total of 6,225 patients (mean age 61.9 years [SD 12.6], 2,284 women [36.5%]) with available primary outcome data were included in these analyses. Overall, only the control of BP and blood glucose contributed positively to the beneficial effect, with mediated proportions of 8.9% (95% CI 4.8–20.0) and 7.0% (1.1–17.0) for achieved systolic BP and blood glucose over 24 hours after randomization, respectively, and 4.0% (1.2–14.0) and 7.6% (2.2–15.0) for reaching the specified targets for systolic BP and blood glucose, respectively. Discussion The major contributors to the effectiveness of the care bundle in INTERACT3 for improved functional outcomes after acute ICH were the control of systolic BP and blood glucose, as indicated by both the achieved levels and protocol targets being met.
 (acceso libre)	Article 31. Moyamoya Disease in Latin America: A Case Report with Systematic Review and Meta-Analysis (P8-13.010) Fritz Fidel Váscones-Román, Brighth Avila Lucas, Marcelo Barriga, Niels Pacheco, and Frank G. Solis Chucos <u>INFO & AFFILIATIONS</u> Neurology Journals, April 8, 2025 Abstract Objective To examine the clinical characteristics, diagnostic approaches, and treatment outcomes of Moyamoya disease (MMD) in Latin America through a case report and a systematic review and meta-analysis. Background Moyamoya disease is a rare cerebrovascular disorder involving progressive stenosis of the internal carotid arteries and abnormal collateral vessel formation. While most prevalent in East Asia, MMD also affects other populations, including those in Latin America, where its presentation and outcomes are less well understood. Design/Methods We conducted a systematic review of Latin American MMD cases, following PRISMA guidelines. Databases searched included PubMed, Embase, and Scopus. Case reports, case series, and cohort studies were analyzed. Data extraction focused on patient demographics, clinical presentation, diagnostic tools, treatments, and outcomes. Meta-analysis was performed, and the risk of bias was assessed using the Joanna Briggs Institute checklist. Results Twenty-one studies, involving 32 patients from Brazil, Mexico, Colombia, Cuba, and Peru, were included. Ischemic strokes were the most common presentation (78%), followed by hemorrhagic events (22%). Digital subtraction angiography (DSA) was the preferred diagnostic method. Treatments varied from medical management to surgical revascularization, such as encephaloduroarteriosynangiosis (EDAS). Meta-analysis showed 40% of patients experienced neurological sequelae, while 78% had favorable outcomes, especially with surgical interventions. Conclusions Moyamoya disease in Latin America predominantly manifests as ischemic events, with DSA as the primary diagnostic tool. Surgical revascularization appears to result in better recovery compared to medical treatment alone. However, due to limited data, further research is needed to better understand MMD in this region and improve treatment strategies.

Preprint

32. Multi-ancestry Transcriptome-Wide Association Study Reveals Shared and Population-Specific Genetic Effects in Alzheimer's DiseaseXinyu Sun^{1,3}, Makaela Mews^{2,3}, Nicholas R. Wheeler^{1,3}, Penelope Benchek^{1,3, 4}Tianjie Gu^{14,15}, Lissette Gomez^{14,15}, Nicholas Ray^{4,5,6}, Christiane Reitz^{4,5,6}, Adam C. Naj^{7,8}, Jennifer Elizabeth Below⁹, Giuseppe Tosto^{4,5,6}, Mario Comejo-Olivas^{10,11}, Goldie S. Byrd¹², Briseida E. Feliciano-Astacio¹³, Katrina Celis^{14,15}, Farid Rajabli^{14,15}, Brian W. Kunkle^{14,15}, Margaret A. Pericak-Vance^{14,15}, Jonathan L. Haines^{1,3}, Anthony J. Griswold^{14,15}, and William S. Bush^{1,3}.¹ Department of Population and Quantitative Health Sciences, School of Medicine, Case Western Reserve University, Cleveland, Ohio, USA.² System Biology & Bioinformatics; Department of Nutrition, School of Medicine, Case Western Reserve University, Cleveland, Ohio, USA.³ Cleveland Institute for Computational Biology, Case Western Reserve University, Cleveland, Ohio, USA.⁴ Taub Institute for Research on Alzheimer's Disease and the Aging Brain, Vagelos College of Physicians and Surgeons, Columbia University, New York, New York, USA.⁵ The Gertrude H. Sergievsky Center, Vagelos College of Physicians and Surgeons, Columbia University, New York, New York, USA.⁶ Department of Neurology, Vagelos College of Physicians and Surgeons, Columbia University, and the New York Presbyterian Hospital, New York, New York, USA.⁷ Department of Biostatistics and Epidemiology, University of Pennsylvania Perelman School of Medicine, Philadelphia, Pennsylvania, USA.⁸ Penn Neurodegeneration Genomics Center, Department of Pathology and Laboratory Medicine, Perelman School of Medicine, University of Pennsylvania, Philadelphia, Pennsylvania, USA.⁹ Vanderbilt University Medical Center.¹⁰ Neurogenetics Working Group, Universidad Científica del Sur, Lima, Peru.¹¹ Neurogenetics Research Center, Instituto Nacional de Ciencias Neurológicas, Lima, Peru.¹² Maya Angelou Center for Health Equity, Wake Forest University, Winston-Salem, North Carolina, USA.¹³ Universidad Central del Caribe, Bayamón, Puerto Rico.¹⁴ John P. Hussman Institute for Human Genomics, Miller School of Medicine, University of Miami, Miami, Florida, USA.¹⁵ Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami Miller School of Medicine, Miami, Florida, USA

bioRxiv, 2025

Abstract

Alzheimer's disease (AD) risk differs across ancestral populations, yet most genetic studies have focused on Non-Hispanic White (NHW) cohorts. We conducted a multi-population transcriptome-wide association study (TWAS) using whole-blood RNA-seq and genotype data from reported NHW (n=235), African American (AA; n=224), and Hispanic (HISP; n=292) participants in MAGENTA. Using SuShiE for multi-population fine-mapping, we identified credible sets of eQTLs for 8,748 genes and improved fine-mapping precision relative to analyses using fewer populations. eQTL effects were largely shared across populations, with population-specific regulation for a subset of genes. Population-stratified TWAS and sample size-weighted meta-analysis (FUSION + MAFOCUS) prioritized and fine-mapped nine genes (FDR<0.05, PIP>0.8), including established AD loci (BIN1, PTK2B, DMPK) with consistent effects across populations. Importantly, at BIN1 we fine-mapped regulatory variants associated with gene expression and AD risk beyond the GWAS index SNP—most notably rs11682128, which is only in modest LD with rs6733839 ($r^2 \approx 0.34$)—demonstrating that multi-population TWAS can implicate additional functional variants not captured by single-SNP GWAS signals. We also discovered a novel association between COG4 expression and AD in NHW, implicating Golgi apparatus function. Using independent SuShiE-derived models from TOPMed MESA (PBM), several associations replicated directionally across ancestries, with statistical significance most evident in NHW. Our results show that multi-population fine-mapping improves eQTL resolution and TWAS interpretability, reveals regulatory variants beyond GWAS index SNPs, and underscores the need to expand non-European AD cohorts to resolve shared and population-specific mechanisms.

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Multi-ancestry Transcriptome-Wide Association Study Reveals Shared and Population-Specific Genetic Effects in Alzheimer's Disease

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Posters presentation

33. Multi-Ethnic Analysis of Expression Quantitative Trait Loci (eQTL) in Alzheimer's Disease: A Focus on Health Disparity Populations

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Alzheimer's & Dementia, 2025 Jan 3;20(Suppl 1)

Abstract**Background**

Despite its high heritability, the genetic mechanisms influencing Alzheimer's Disease (AD), particularly in health disparity populations like African Americans (AA) and Hispanics (HI), are not fully understood. The lack of ancestral diversity in genetic datasets, notably in eQTL studies that associate genetic variation with gene expression, exacerbates these disparities. Our study seeks to address this gap by comparing the AD interactions of racially and ethnically diverse expression Quantitative Trait Loci (eQTL) effects to investigate the genetic influence on AD in underrepresented populations.

Method

We investigated the impact of AD status on multi-ethnic eQTLs across 3 diverse ancestral cohorts: AA, HI, and Non-Hispanic White (NHW). Genotype and gene expression data were collected from samples of each genetic ancestry (AA = 224, HI = 293, NHW = 235), all with known AD status, with a focus on generating whole blood RNAseq data. We modified the TensorQTL pipeline to incorporate an interaction term for AD status to identify eQTL effects potentially induced by disease. We examined the top eQTL association per gene to assess the impact of AD status across the three ancestral cohorts.

Result

Our preliminary analysis reveals striking variations in the impact of AD status on gene expression across AA, HI, and NHW cohorts. Several genes in each ancestry show statistically significant ($p < 10^{-8}$) interaction effects (G X AD) (AA = 11, HI = 5, NHW = 9), indicating that genetic variation within those genes influences gene expression in an AD-status dependent manner. A number of genes displayed interesting interaction patterns, where the most significant eQTL-AD interaction was identical in two of the three ancestries studied, suggesting a potential ancestrally-shared connection to AD risk (AA+HI = 4, AA+NHW = 3, HI+NHW = 5, AA+HI+NHW = 0), though notably there were no genes with induced eQTLs across all three ancestries.

Conclusion

Our study highlights the impact the AD disease process has on genetic variants that alter whole-blood gene expression. We also highlight differences by population, prompting the development of ethnically diverse gene expression panels that pave the way for more accurate genetic studies in health disparity populations. Our findings also underscore the potential for discovering novel genetic mechanisms influencing AD risk, which could lead to more effective and inclusive therapeutic strategies.



Article

34. Multiple molecular diagnoses identified through genome sequencing in individuals with suspected rare disease

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HGG Advances, Volume 6, Issue 3, 2025

Summary

Genome sequencing is a powerful and comprehensive test that detects multiple variants of different types. The interrogation of variants across the genome enables the identification of multiple molecular diagnoses (MMDs) in a single individual. In this retrospective study, we describe individuals in whom MMDs were associated with the proband's indication for testing (IFT), secondary findings, or incidental findings. An MMD is considered where at least one of the findings is associated with the primary IFT and all variants are classified as either likely pathogenic or pathogenic. Clinical genome sequencing was performed for all individuals as part of the iHope program at the Illumina Laboratory Services between September 2017 and December 2023. The iHope cohort included 1,846 affected individuals, with 872 (47.2%) found to have at least one likely pathogenic or pathogenic variant associated with the primary IFT. Of these, 81 (9.3%) individuals had multiple clinically significant molecular findings, including 76 individuals with reported variants associated with 2 different conditions, and 5 individuals with more than 2 molecular findings. A total of 32 individuals (3.7%) had at least 2 molecular diagnoses related to the primary IFT, while in 49 (5.6%) individuals, the variant(s) reported for the second condition constituted a secondary or incidental finding. Our study highlights that among individuals with a likely pathogenic or pathogenic finding identified through genome sequencing, 9% have MMDs, which may have been missed with different testing methods. Of note, approximately 60% of the 81 individuals with an MMD had a potentially actionable secondary or incidental finding.

Brief communication

35. Myelin-oligodendrocyte glycoprotein (MOG) antibody-associated acute disseminated encephalomyelitis (ADEM) in an adult with dengue

Leonardo Torioni, Isidro Gonzales Quispe, Michel Elyas Jung Haziot, Mariana Vaz-Curado, Rene L. M. Rivero & José Ernesto Vidal

Neurological Sciences, Volume 46, 2025

Abstract

Dengue is the most worldwide arbovirus and uncommonly causes encephalitis or acute disseminated encephalomyelitis (ADEM). Myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD) has recently been recognized as a spectrum of monophasic or relapsing course demyelinating syndromes. We present the first case of MOGAD ADEM in an adult with dengue from Brazil. The patient was a 21-year-old man with no relevant medical or epidemiological history and a confirmed diagnosis of dengue with no warning

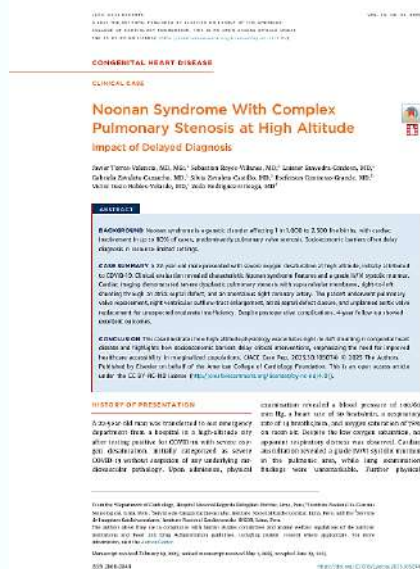


(accesso libre)



(suscripción o compra)

signs after three days of classical clinical manifestations. In the next days, the patient had lower limb weakness, acute urinary retention, diplopia, sonolence, and mental confusion. MRI showed multiple cerebral and spinal cord lesions compatible with ADEM. CSF analysis demonstrated pleocytosis (135 WBC/μL) and elevated protein (155 mg/L). Dengue-IgM antibodies were positive in the CSF. Serum MOG-IgG cell-based immunofluorescence assay was positive with a titer >1:160. The patient received methylprednisolone for five days, showing progressive and unequivocal improvement of neurological manifestations and he was discharged with oral prednisone. This report suggests an emerging association between dengue-induced immune dysregulation and the development of MOGAD-associated ADEM. The presence of MOG antibodies should be considered in adults with dengue-related ADEM and timely syndromic management may determine favorable outcomes.



(acceso libre)

Clinical case

36. Noonan Syndrome With Complex Pulmonary Stenosis at High Altitude: Impact of Delayed Diagnosis

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JACC: Case Reports, Volume 30, Issue 28, 17 September 2025

Abstract

Background

Noonan syndrome is a genetic disorder affecting 1 in 1,000 to 2,500 live births, with cardiac involvement in up to 80% of cases, predominantly pulmonary valve stenosis. Socioeconomic barriers often delay diagnosis in resource-limited settings.

Case Summary

A 22-year-old male presented with severe oxygen desaturation at high altitude, initially attributed to COVID-19. Clinical evaluation revealed characteristic Noonan syndrome features and a grade IV/VI systolic murmur. Cardiac imaging demonstrated severe dysplastic pulmonary stenosis with supravalvular membrane, right-to-left shunting through an atrial septal defect, and an anomalous right coronary artery. The patient underwent pulmonary valve replacement, right ventricular outflow-tract enlargement, atrial septal defect closure, and unplanned aortic valve replacement for unexpected moderate insufficiency. Despite postoperative complications, 4-year follow-up showed excellent outcomes.

Conclusion

This case illustrates how high-altitude physiology exacerbates right-to-left shunting in congenital heart disease and highlights how socioeconomic barriers delay critical interventions, emphasizing the need for improved healthcare accessibility in marginalized populations.



(acceso libre)

Clinical/Scientific note

37. Novel ATXN10 Repeat Motif Patterns in Peruvian Families Modify Disease Onset

Kamilla Sedov, Carla Manrique-Enciso, Madison James Yang, Ismael Araujo-Aliaga, Egor Dolzhenko, Samantha Kalla, Sarah Bowman Kingan, Elison Sarapura-Castro, Andrea Rivera-Valdivia, Maryenela Zaida Illanes-Manrique, Mario Cornejo-Olivas, and Birgitt Schüle. [Authors info & affiliations.](#)

Neurol Genet, 2025

Abstract**Objectives**

Spinocerebellar ataxia type 10 (SCA10) is an autosomal-dominant disorder caused by intronic expansions of pentanucleotide repeats in the ATXN10 gene. While various repeat motifs have been described, emerging evidence suggests that specific repeat motifs, rather than repeat length alone, can modify disease features such as seizure prevalence and penetrance.

Methods

We used a novel multiplex 20-gene panel with Cas9-targeted, amplification-free long-read sequencing (LRS) and optical genome mapping to elucidate ATXN10 repeat motif patterns and investigated genotype-phenotype correlations in index cases of 6 multigenerational SCA10 kindreds from Peru.

Results

We detected ATXN10 repeat expansions ranging from 990 to 2,002 pentanucleotide repeats (4.9–10 kb) across 6 families. It is important to note that we identified 3 mixed repeat motif patterns and ratios of (ATTCT)_n(ATTCC)_m, which were associated with differences in age at disease onset and anticipation.

Discussion

Our key novel finding is the prominence of the alternate ATTCC motif alongside the common ATTCT motif. While repeat length alone does not appear to drive disease onset in SCA10, we uncovered that the ratio of the ATTCC motif within distinct repeat patterns might correlate with disease onset. These findings underscore the need to adapt LRS clinical workflows to fully characterize large repeat expansions at the nucleotide level.



(suscripción o compra)

Article

38. Novel Intermediate ATXN10 Alleles in the Healthy Peruvian Population: A Matter of Indigenous American Ethnic Origin

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#Contributed equally.

Cerebellum. 2025 Feb 7;24(2)

Abstract

Spinocerebellar ataxia type 10 (SCA10) is a neurodegenerative disease predominant in Latin American individuals with Indigenous American ancestry. SCA10 is caused by an expansion of ATTCT repeat within the ATXN10 gene. Healthy individuals carry 9-32 ATTCT repeats, whereas SCA10 patients carry an expansion of 280 repeats and higher. Recently, intermediate alleles (over than 32 repeats) have been identified in healthy Peruvian Indigenous American individuals, with unclear significance. This study aims to characterize the variability of the ATTCT repeats within the ATXN10 gene across self-declared Indigenous American and Mestizo subpopulations from Peru. A total of 871 samples (754 Mestizo and 117 Indigenous American) were analyzed using PCR, and RP-PCR when suspecting apparent homozygosity due to larger alleles. 8.7% of the total of healthy individuals (76/871) carry at least one intermediate allele. The 14-repeat allele being the most common for both subpopulations (41.5%). Intermediate alleles were detected in the Peruvian population (4.5%) with a significantly higher frequency among self-declared Indigenous American compared to Mestizo, suggesting a possible association with the ethnic origin. The G allele at the SNP rs41524547 had a frequency of 51.39% in individuals with intermediate alleles, with not significantly difference between subpopulations. Further analysis should be performed to confirm the size and composition of ATTCT repeat tract, as well as the contribution of rs41524547 in SCA10.

Article

39. Performance of skimmed milk flocculation and real-time polymerase chain reaction for the concentration, detection, and quantification of *Taenia solium* in urban wastewater from an endemic region of the Peruvian central highlands

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Science of The Total Environment, Volume 977, 15 May 2025

Abstract

The taeniasis/cysticercosis complex, caused by *Taenia solium*, is a significant zoonotic disease in low- and middle-income countries. Although this complex has been extensively studied in rural communities, information on its status in urban areas remains limited due to the financial and logistical challenges of conducting large-scale assessments in cities. This study aimed to demonstrate the feasibility of using wastewater monitoring for the epidemiological surveillance of *T. solium* in cities. We evaluated four extraction methods (magnetic beads, bead-beating, spin column, and thermal shock) on serial dilutions of *T. solium* eggs. Statistical analysis demonstrated that the magnetic bead method achieved the highest quantifications of genomic copies (GC). These values were used to develop a simple linear regression to establish an equivalence between the GC of the internal transcribed spacer 1 (ITS-1) region and eggs. Subsequently, we assessed the performance of skimmed milk



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Performance of skimmed milk flocculation and real-time polymerase chain reaction for the concentration, detection, and quantification of *Taenia solium* in urban wastewater from an endemic region of the Peruvian central highlands

Cesar A. Valdivia-Carrera ^{a b}, Ana C. Ho-Palma ^c, Luis A. Gomez-Puerta ^d, Daniel A. Andrade ^{a b}, Brenda L. Gonzales ^{a b}, Maria T. Lopez-Urbina ^e, Armando E. Gonzalez ^d, Javier A. Bustos ^{f g}, Hector H. Garcia ^{f g h}, Sarah Gabriël ⁱ, Seth E. O'Neal ^{g i}, Eloy Gonzales-Gustavson ^{a b}

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Highlights

- Monitoring of urban wastewater revealed the presence of *Taenia solium* DNA and eggs.
- Wastewater monitoring could be a community surveillance tool for *T. solium*.
- Skimmed milk flocculation allowed *T. solium* eggs to be concentrated in wastewater.
- The magnetic bead method yielded high DNA extraction from *T. solium* eggs.



(suscripción o compra)

flocculation, along with the impact of inhibitory substances, in two distilled water samples and two wastewater samples (250 mL each) seeded with low (5000) and high (25000) eggs numbers. Recovery percentages in distilled water were $98 \pm 6\%$ (high concentration) and $26 \pm 11\%$ (low concentration), while in wastewater, they were $76 \pm 11\%$ and $12 \pm 3\%$, respectively. Over 16 months, wastewater monitoring in a highly endemic city of the Peruvian Central Highlands detected *T. solium* in 17 % (12/70) of samples using a real-time polymerase chain reaction targeting the *T. solium* ITS-1 region, with concentrations ranging from $8.92E+02$ to $8.07E+06$ GC/100 mL, equivalent to 0.7 and 4510 eggs/100 mL, respectively. Sanger sequencing of the cytochrome c oxidase subunit 1 gene confirmed the presence of *T. solium* in 4 of the 12 positive samples for the ITS-1 region. In conclusion, *T. solium* can be detected and quantified in wastewater, supporting its use for urban human taeniasis surveillance.

Poster presentation

40. "Polygenic Genetic Risk Factors in Alzheimer's Disease: A Case-Control Study in Peruvian Population"

Jonathan Adrian Zegarra-Valdivia^{1,2,3}, Harold Alessandro Arana-Nombera¹, Leandro Aron Perez-Fernandez¹, Milagros del Rocio Casimiro-Arana¹, Reyna Elizabeth Alamo-Medina¹, Viviana Nayelli Gallegos-Manayay^{1,2}, María del Rosario Oliva-Piscoya¹, Nobuko Vásquez-Zuñe¹, Eduardo Sebastián Abanto-Saldaña¹, María Celinda Cruz Ordinola¹, Carmen Paredes Manrique³, Miguel E Renteria^{4,5}, Sheila Castro-Suarez^{6,7}; Neuroscience and Mental Health Group

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Alzheimer's & Dementia. 2025 Jan 3;20(Suppl 1)

Abstract

Background

Alzheimer's Disease (AD) is the most prevalent form of dementia globally. While some familial cases are observed, sporadic AD cases are more common and reflect a high level of complexity, with individual risk determined by the interaction of polygenic and environmental factors.

Objective

To characterize polygenic genetic risk factors in individuals with cognitive impairment and Alzheimer's Disease across four regions of Peru.

Method

This will be a prospective observational case-control study. We will identify the neurocognitive profile of participants and collect saliva samples for subsequent genetic profiling. Participants will be divided into three main groups: those with cognitive impairment, those diagnosed with AD, and control subjects over 50 years old without dementia.

Results

We expect to validate the effect of risk variants previously identified in the European population within the Peruvian population. Additionally, the aim is to identify new genetic risk factors specific to the Peruvian population. Collectively, the findings from this study could lay the groundwork for implementing a polygenic risk score model, which would estimate an individual's genetic risk of developing Alzheimer's Disease.

Alzheimer's & Dementia
THE JOURNAL OF THE ALZHEIMER SOCIETY

"Polygenic Genetic Risk Factors in Alzheimer's Disease: A Case-Control Study in Peruvian Population"

Jonathan Adrian Zegarra-Valdivia^{1,2,3} | Harold Alessandro Arana-Nombera¹ |
Leandro Aron Perez-Fernandez¹ | Milagros del Rocio Casimiro-Arana¹ |
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María del Rosario Oliva-Piscoya¹ | Nobuko Vásquez-Zuñe¹ |
Eduardo Sebastián Abanto-Saldaña¹ | María Celinda Cruz Ordinola¹ |
Carmen Paredes Manrique³ | Miguel E. Renteria^{4,5} | Sheila Castro-Suarez^{6,7} |
Neuroscience and Mental Health Group

Abstract
Background: Alzheimer's Disease (AD) is the most prevalent form of dementia globally. While some familial cases are observed, sporadic AD cases are more common and reflect a high level of complexity, with individual risk determined by the interaction of polygenic and environmental factors.
Objective: To characterize polygenic genetic risk factors in individuals with cognitive impairment and Alzheimer's Disease across four regions of Peru.
Method: This will be a prospective observational case-control study. We will identify the neurocognitive profile of participants and collect saliva samples for subsequent genetic profiling. Participants will be divided into three main groups: those with cognitive impairment, those diagnosed with AD, and control subjects over 50 years old without dementia.
Results: We expect to validate the effect of risk variants previously identified in the European population within the Peruvian population. Additionally, the aim is to identify new genetic risk factors specific to the Peruvian population. Collectively, the findings from this study could lay the groundwork for implementing a polygenic risk score model, which would estimate an individual's genetic risk of developing Alzheimer's Disease.

Keywords: Alzheimer's Disease, Polygenic Risk Factors, Case-Control Study, Peruvian Population, Cognitive Impairment, Alzheimer's Disease.

Alzheimer's & Dementia 2025 Jan 3;20(Suppl 1)
doi:10.1002/alz.12345



(acceso libre)

Conclusion

This study aims to significantly contribute to the understanding of Alzheimer's Disease's genetic underpinnings in the Peruvian population. By exploring both known and novel genetic risk factors, we hope to enhance early detection strategies and personalized risk assessments for AD, ultimately guiding more targeted interventions and preventative measures.

Original research article

41. Prevalence and risk factors of mild cognitive impairment and dementia in northern Peru

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Front Public Health, 20 May 2025

Abstract

Background

Global dementia prevalence estimates indicate a growing burden, particularly in Low- and Middle-Income Countries (LMICs). Factors such as education, socioeconomic status, and limited public health interventions contribute to the development of Alzheimer's disease and other dementias. This study aimed to determine the prevalence of Mild Cognitive Impairment (MCI) and dementia in middle-aged and older adults from northern Peru, as well as identify possible associated risk factors.

Methods

A cross-sectional study was conducted with 385 participants aged 40 to 85 years from Chiclayo, Peru. Cognitive impairment was assessed using the Rowland Universal Dementia Assessment Scale (RUDAS) and INECO Frontal Screening (IFS). Functional activity and depression were evaluated with the Pfeffer Functional Activities Questionnaire (PFAQ) and Patient Health Questionnaire-9 (PHQ-9). Participants were classified as controls, MCI, or dementia based on education-adjusted cutoff scores of those scales through cognitive system classification tools.

Results

According to the results, 31.4% of the sample consisted of subjects without cognitive impairment, 40.5% were identified as possible cases of MCI, and 24.9% as possible cases of dementia. However, the percentages by age group are high. A lower educational level is associated with older age and correlates with lower cognitive scores and functional impairment. Age, hypertension, and hearing loss were significant risk factors for MCI and dementia.

Conclusion

The prevalence of possible MCI and dementia in a city in north Peru is high, with a predominance of MCI and dementia in older adults. Age, low education, hypertension, and hearing loss are potential risk factors for cognitive impairment.



(acceso libre)



RESEARCH ARTICLE

Prevalence of Parkinson's Disease in a Town of the Central Highlands of Peru: A Population-Based Study

Elison Sarapura-Castro MD, Karina Milla-Nevra BS, Julia Ríos-Pinto MSc, Jorge L. Maguina MSc, Andrea Rivera-Valdivia MD, Miguel Inca-Martínez BS, Maryenela Illanes-Manrique MD, Milagros Galecio-Castillo MD, Silvia Montano MD, MPH, Ignacio F. Mata PhD, Mario Comejo-Olivas MD

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Abstract**Background**

The global prevalence of Parkinson's disease (PD) is estimated to be 315 per 100,000 inhabitants, with variations across regions and depending on the epidemiological methods used. Door-to-door prevalence studies in Andean communities are notably underrepresented in the literature.

Objective

The aim of this study was to estimate the PD prevalence in Jauja.

Methods

A cross-sectional community-based door-to-door prevalence study was conducted from April 2018 to September 2019. We used a Spanish version of the 9-item screening questionnaire (9-item SQ). Trained interviewers conducted face-to-face screening surveys. Participants presenting "suspected parkinsonism" underwent further neurological assessment. PD cases responded to the LANGE-PD questionnaire and "pesticide exposure questionnaire." We calculated the crude prevalence, gender- and age-specific rates, and age-adjusted rates.

Results

Out of 2527 eligible individuals, 1960 completed the screening survey. Of these, 563 were selected for neurological assessment, and 337 underwent neurological examination. PD was confirmed in 21 individuals, 16 of whom were identified using the 9-item SQ with a crude prevalence of 816/100,000. We observed higher rates of PD in older age groups and in males compared to females (1.6 vs. 0.3%, $P < 0.05$). The age-standardized prevalence was 566/100,000 (95% confidence interval [CI]: 496.6–635.2). Among PD cases, the most common environmental exposures were living near farming fields and pesticide exposure.



(suscripción o compra)

Research article

42. Prevalence of Parkinson's Disease in a Town of the Central Highlands of Peru: A Population-Based Study

Elison Sarapura-Castro MD, Karina Milla-Nevra BS, Julia Ríos-Pinto MSc, Jorge L. Maguina MSc, Andrea Rivera-Valdivia MD, Miguel Inca-Martínez BS, Maryenela Illanes-Manrique MD, Milagros Galecio-Castillo MD, Silvia Montano MD, MPH, Ignacio F. Mata PhD, Mario Comejo-Olivas MD

Movement Disorders Clinical Practice / Volume 13, Issue 3, 2026

Abstract**Background**

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Out of 2527 eligible individuals, 1960 completed the screening survey. Of these, 563 were selected for neurological assessment, and 337 underwent neurological examination. PD was confirmed in 21 individuals, 16 of whom were identified using the 9-item SQ with a crude prevalence of 816/100,000. We observed higher rates of PD in older age groups and in males compared to females (1.6 vs. 0.3%, $P < 0.05$). The age-standardized prevalence was 566/100,000 (95% confidence interval [CI]: 496.6–635.2). Among PD cases, the most common environmental exposures were living near farming fields and pesticide exposure.

Conclusions

The observed prevalence of PD in this town in the central highlands of Peru aligns with previous reports, with notably higher rates among individuals over the age of 80. Further prospective studies are warranted to investigate the potential influence of environmental factors on PD prevalence in this region.



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(suscripción o compra)

Short report

43. Quantification of Anti-GP50, Anti-rT24H, and Anti-sTs18var1 Antibodies to Identify Viable Infection in Patients with Neurocysticercosis

Andrea Rivera, Luz M. Toribio, Javier A. Bustos, Herbert Saavedra, Isidro Gonzales and Héctor H. García for The Cysticercosis Working Group in Peru

Am. J. Trop. Med. Hyg., Volume 114, 2025

Abstract

Identifying viable infections in neurocysticercosis (NCC) is crucial for treatment. Neuroimaging is the primary diagnostic tool, but it is not widely available. Moreover, in many cases, imaging diagnosis is not pathognomonic and requires serological confirmation. The serological assay of choice, enzyme-linked immunoelectrotransfer blot using lentil lectin-purified glycoprotein (LLGP-EITB) *Taenia solium* antigens to detect specific antibodies, exhibits high predictive values for the presence of viable NCC when the results are positive for multiple (>3) antibody bands; it also exhibits high predictive values for the absence of viable infection when the results are negative or the test reacts to a single

antibody band. However, its interpretation in terms of viable infection is limited in cases with two or three positive bands (intermediate results), which occur in one-quarter of patients with NCC. The quantification of specific antibodies could allow for the identification of viable infections. Using a multi-antigen, quantitative multiplex bead assay, antibody levels were measured against *Taenia solium* proteins rGP50, rT24H, and sTs18var1 in 94 patients with intermediate LLGP-EITB results. The antibody-to-rT24H (25.96 versus 5.49; $P = 0.0048$) and antibody-to-sTs18var1 ratios (3.62 versus 1.37; $P = 0.0083$) were higher in subjects with viable cysticerci than in controls. Patients with high antibody levels against the proteins rT24H and sTs18var1 were 5.4 times more likely to have a viable infection than those with low antibody levels. The quantification of antibodies against rT24H and sTs18var1 can help define a viable NCC infection.

Article

44. Risk and impact of stroke across 38 countries and territories of the Americas from 1990 to 2021: a population-based trends analysis from the Global Burden of Disease Study 2021

Ramón Martínez ^a, Paula Muñoz-

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Fabián Alvarez ^g, Pablo Amaya ^h, Sebastian Ameriso ^e, Antonio Arauz ⁱ, Miguel A. Barboza ^j, Hernán Bayona ^{k l}, Antonio Bernabé-

Ortiz ^m, Juan Calleja ⁱ, Vanessa Cano-Nigenda ⁱ, Leonardo

Augusto Carbonera ⁿ, Rodrigo M. Carrillo-Larco ^o, Angel Corredor ^p, Ana Cláudia de

Souza ⁿ, Claudio Jimenez ^l, Fernando Lanas ^q, Sheila Martins ^{n r s}, Víctor Navia ^{t u}, Nelson Navarro-

Escudero ^v, Verónica Olavarria ^t, Bruce Ovbiagele ^w, Kevin Pacheco-Barrios ^x, Octavio Pontes-

Neto ^y, Virginia Pujol ^e, Alejandro Rabinstein ^z, Julieta Rosales ^e, Andrés Rose ^{nde a}, Gisele Sampaio ^{silva aa}, Gustavo Saposnik ^{ab}, Souvik Sen ^{ac}, Fernando D. Testai ^{ad}, Victor Urrutia ^{ae}, Craig S. Anderson ^{af ag}, Pablo M. Lavados ^t

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(Continúa)

The Lancet Regional Health – Americas, Vol 43, 2025

Summary

Background

Despite substantial declines in burden over time, stroke remains a public health threat in the Americas. This study aimed to assess the current magnitude, trends, and disparities in the estimates of stroke burden by sex and age in the Americas from 1990 to 2021.

Methods

Estimates from the *Global Burden of Disease, Injuries and Risk Factors Study 2021* were used to analyze incidence, prevalence, mortality, years of life lost due to premature death, years lived with disabilities, and disability-adjusted life years (DALYs) caused by stroke and its major subtypes stratified by age, and sex in the Americas from 1990 to 2021. We used Joinpoint regression analysis to estimate the average annual percent change (AAPC) of stroke mortality and disease burden outcomes and assessed trends.



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	Findings In 2021, there were 1.1 million (95% uncertainty interval: 1.0–1.2) new cases, 12.9 million (12.3–13.7) prevalent cases, 0.5 million (0.5–0.6) deaths, and 11.4 million (10.6–12.1) DALYs due to stroke in the Americas. The absolute number of stroke burden outcomes increased from 1990 to 2021, but their corresponding age-standardized rates significantly declined. A deceleration in reduction rates of burden outcomes for all strokes and most stroke subtypes occurred over the last decade, with pronounced difference between sexes mainly in incidence among younger groups. From 2015 to 2021, trends in incidence rates from all stroke and stroke subtypes reversed to increase in most age groups, and strikingly, trends in mortality and DALY rates from <u>ischemic stroke</u> among younger populations reversed to upward with AAPC over 1.4%. A substantial number of countries contributed to these increasing trends. Interpretation Regionally, the annual number of stroke cases and deaths significantly increased from 1990 to 2021, despite reductions in age-standardized rates. The declining pace in age-standardized stroke rates has decelerated in recent years, while trends in incidence, and <u>ischemic stroke</u> mortality and DALY among middle-aged adults and adults, reversed towards upward in the period 2015–2021. Further studies are needed to understand the determinants of this recent pattern and identify the most cost-effective interventions to stem this alarming trend. Funding There was no funding source for this study.
 The screenshot shows the title page of the article. It includes the journal name 'Parkinsonism & Related Disorders', volume 134, May 2025, pages 107944. The title is 'Sex differences in the diagnosis latency of Parkinson's disease in Latin America'. The authors listed are Janvi Ramchandra, Miguel Inca-Martinez, Thiago Peixoto Leal, Henry Mauricio Chaparro-Solano, Amira Salim, Emilia M. Gatto, Natalia González Rojas, Gustavo Da Prat, Federico Micheli, Bruno Lopes Santos-Lobato, Francisco E.C. Cardoso, Sarah Camargos, Grace H. Letro, Pedro Braga-Neto, Vitória Maria Torres Peixoto, Artur F.S. Schuh, Vitor Tumas, Manuelina M. Brito, Vanderici Borges, Carolina Candeias da Silva, and Ignacio F. Mata. There are highlights at the bottom: 'Overall sex differences in PD age at diagnosis were found.', 'Sex differences in PD age at onset were found in Mexico.', 'Sex differences in PD diagnosis latency exist in Chile & diagnoses at <39 & >70.', and 'Age at onset in our Latin American cohort is ~6 years younger than Europeans.'	Article 45. Sex differences in the diagnosis latency of Parkinson's disease in Latin America Janvi Ramchandra a b, Miguel Inca-Martinez a, Thiago Peixoto Leal a, Henry Mauricio Chaparro-Solano a c, Amira Salim a c, Emilia M. Gatto d, Natalia González Rojas e, Gustavo Da Prat e, Federico Micheli f g, Bruno Lopes Santos-Lobato h i, Francisco E.C. Cardoso j, Sarah Camargos j, Grace H. Letro k, Pedro Braga-Neto l m, Vitória Maria Torres Peixoto l, Artur F.S. Schuh n o, Vitor Tumas p, Manuelina M. Brito p, Vanderici Borges q, Carolina Candeias da Silva q, Henrique B. Ferraz q, Pedro Chana-Cuevas r, Paula Saffie-Awad s r t, Patricio Olguin u v, Alicia Colombo w x, Andres de la Cerda y, Gonzalo A. Farias z aa, Juan Cristobal Nuñez aa, Gonzalo Arboleda ab, Humberto Arboleda ac, William Fernandez ab, Carlos E. Arboleda-Bustos ac, Jorge L. Orozco ad ae, Beatriz Muñoz-Ospina af, Carlos Velez-Pardo ag, Marlene Jiménez-Del-Río ag, Francisco Lopera ag, Sonia Moreno ag, David A. Pineda ag, Omar Buritica ag, Gabriel Torrealba-Acosta ah ai, Alex Medina Escobar aj, Mayela Rodríguez-Violante ak, Ana Jimena Hernández-Medrano al, Daniel Martínez-Ramírez am, Mirna González-González am, Miguel E. Rentería an, Sarael Alcauter ao, Paula Reyes-Pérez ap, Alejandra Medina-Rivera ap, Damaris Vazquez-Guevara aq, Lucero de María Ugalde-Mejía ar, Maritza J. Valadez M. as, Omar Cárdenas-Sáenz at, Ildefonso Rodríguez-Leyva au av, Carlos M. Guerra-Galicia aw, Nadia A. Gandarilla-Martínez ax, Yamil Matuk-Pérez ay az, Eugenia Morelos-Figaredo ba, Karla Salinas-Barboza bb, Sara Isais-Millán bc, Teresa Pérez-Torres bd, Diana C. Deras Gaucin be, Alejandra E. Ruiz-Contreras bf, Ingrid Estrada-Bellmann bg, Julia Rios-Pinto bh bi, Mario Cornejo-Olivas bj bi, Carlos Cosentino bk bl, Luis Torres Ramirez bk, Nicanor Mori bm, Koni Mejía-Rojas bm bn, Ángel César Medina bo, Iván Cornejo-Herrera bp, Edward M. Ochoa bq, Ángel Viñuela br, Elena Dieguez bs, Ignacio Amorín bs, Andrés Lescano bs, Ignacio F. Mata a c aGenomic Medicine Institute, Cleveland Clinic Foundation Lerner Research Institute, Cleveland, USA bDepartment of Biochemistry, Case Western Reserve University, Cleveland, USA cCleveland Clinic Lerner College of Medicine, Case Western Reserve University, Department of Molecular Medicine, Cleveland, USA dDepartment of Neurology, Affiliated University of Buenos Aires, Buenos Aires, Argentina eDepartment of Neurology, Institute of Neuroscience of Buenos Aires (INEBA), Sanatorio de la Trinidad Mitre, Buenos Aires University, Buenos Aires, Argentina fParkinson's Disease and Movement Disorders Center, University of Buenos Aires, Buenos Aires, Argentina gCentro de Parkinson y Movimientos Anormales, Fundación San Gabriel, Buenos Aires, Argentina hHospital Ophir Loyola, Belém, Brazil iLaboratório de Neuropatologia Experimental, Universidade Federal do Pará, Belém, Brazil

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Parkinsonism & Related Disorders, Volume 134, May 2025

Abstract

Background

Age and sex are known risk factors for Parkinson's Disease (PD), but it remains controversial if there are sex differences in the diagnosis latency. The objective of this study was to examine these sex differences in Latin America.

Methods

The Latin American Research Consortium on the Genetics of PD (LARGE-PD) includes PD patients from countries across Latin America who were diagnosed using the UK Brain Bank criteria. Ages at onset (AAO; N = 2,792), diagnosis (AAD; N = 1,416), and calculated diagnosis latency (N = 1,416) were extracted from the LARGE-PD database and compared for both males and females overall, by country, and decade-long age ranges. A cohort was created based on available data for motor sign at onset (N = 492). Regressions examining diagnosis latency as a factor of sex, country, and motor subtype were performed. Two-tailed t-tests at 95 % confidence intervals were used to identify differences in mean AAOs, AADs, and diagnosis latencies between the sexes.

Results

Across the LARGE-PD cohort, lower AAD was observed in males. Per country, AAO was lower for males in Mexico and diagnosis latency was shorter for males in Chile. Overall, younger females (≤ 39) and older males (≥ 70) are likely to experience longer latencies.

Conclusions

Our results suggest that there may be country and age dependent sex differences in AAO, AAD, and diagnosis latency of PD in Latin America. Interestingly, the mean AAO of LARGE-PD is approximately 6 years younger than studies done with European populations. Analyses with additional data are needed to determine the influence of other factors.

Article

46. Social Determinants of Health in Alzheimer's Disease Patients from Peru, Nested Study From the Peruvian Alzheimer Disease Initiative (PeADI) (P11-3.005)

Maryenela Illanes-Manrique, Pilar Giron-Davila, Midiam Silva-Bullon, Larry Adams, Sheila Castro-Suarez, Julia Rios-Pinto, Pedro Mena, Farid Rajabli, Katalina McInerney, Ivan Cornejo-Herrera, Michael L. Cuccaro, Edward Ochoa-Valle, Jeffery Vance, Angel Medina-Colque, Koni Mejia-Rojas, Elison Sarapura-Castro, Sergio Lannata, Azizi Seixas, Margaret Pericak-Vance, and Mario Cornejo Olivas

AUTHORS INFO & AFFILIATIONS

Neurology Journals, April 8, 2025 issue 104 (7_Supplement_1)

Abstract

Objective

We aim to identify the main SDOH associated with Alzheimer's disease (AD) by comparing AD cases to cognitively unimpaired (CU) individuals in the PeADI study.

Background

Social, behavioral and environmental determinants of health (SDOH) such as access to and quality of education and healthcare, and socioeconomic factors influence health outcomes, including dementia.

Design/Methods

The Social Determinants of Brain Health Questionnaire (SDOBH-Q) underwent expert review by local and international neurologists/psychiatrists and local linguistic adaptation. We administer the revised questionnaire to over 100 AD cases and 100 CU individuals from an ongoing genetic study of AD (PeADI) in



(acceso restringido)

Peru. This includes participants from three predominantly rural regions in the Andean highlands (Huancayo, Cusco, Puno) and three coastal urban regions (Lima, Callao, Tacna). The association between identified SDOH and AD status will be analyzed using multivariable regressions, adjusted for age, gender, and population substructure.

Results

The revised SDOBH-Q questionnaire consists of 15 sections and 79 questions. The pilot assessment was completed by 28 CU volunteers: 17 from Lima and 11 from Huancayo. Preliminary data from the first 60 participants have been collected across regions from both the coast and the Andean highlands of Peru, showing differences between AD and CU regarding neighborhood, education, healthcare access and economic conditions.

Research article

47. Spanish translation, adaptation, and validation of the Epilepsy Surgery Satisfaction Questionnaire-19

Elena Fonseca, Alejandro Thomson, Mario Alonso, Óscar Contreras, Walter de la Cruz, María Garcés-Pellejero, Enric Bellido-Castillo, Camilo Espinosa-Jovel, Alberto Velásquez, Enrique de Font-Reaulx, Cintia Flores, Luis Carlos Mayor-Romero, Paula Martínez, Xiana Rodríguez-Osorio, Álvaro Juiz, Mar Carreño, Estefanía Conde, Marta Olivera, Elliot Barreto-Acevedo, Oswaldo Rodríguez, Juan Rodríguez-Uranga, Iratxe Maestro, Mercè Falip, Guillermo Hernández, José M. Serratos, Esteban Cordero, Manuel Quintana, Samuel López-Maza, Daniel Campos-Fernández, Laura Abaira, Estevo Santamarina, Samuel Wiebe, Manuel Toledo

Epilepsia, october 2025

Abstract

Objective

The effectiveness of surgery in drug-resistant epilepsies is often focused exclusively on seizure control. The Epilepsy Surgery Satisfaction Questionnaire-19 (ESSQ_19), developed in 2020, is a reliable tool for assessing the level of satisfaction of patients undergoing surgery. We aimed to perform a Spanish translation, adaptation, and validation of the ESSQ_19 questionnaire.

Methods

This was a prospective multicenter study (five Spanish-speaking countries) including an international cohort of adult patients who underwent epilepsy surgery at least 1 year prior to participation. We followed a translation and back-translation methodology to obtain the Spanish version of the questionnaire (ESP-ESSQ_19). For validation, internal consistency (Cronbach alpha) and test-retest reliability at 6 months were assessed, and psychometric properties were measured by correlating them with parallel aspects using validated questionnaires.

Results

One hundred fifty patients were included (59.3% women; mean age = 39.3 ± 13 years, interquartile range [IQR] = 28–49 years), of whom 94 completed the baseline and follow-up questionnaires, with a median time since surgery of 3 (IQR = 1–7) years. Temporal lobe epilepsy was the most common ($n = 112$, 77.8%), as was structural etiology ($n = 139$, 95.2%). At inclusion, 101 patients (68.7%) had been seizure-free for ≥ 1 year. The ESP-ESSQ_19 questionnaire showed an internal consistency index of .88–.95 for all domains and a test-retest reliability of .91 (95% confidence interval = .87–.94). Significant correlation coefficients were observed between the ESP-ESSQ_19 questionnaire and all validated questionnaires used.

Significance

The ESP-ESSQ_19 questionnaire is a Spanish-language instrument useful for assessing patient satisfaction after epilepsy surgery, with adequate psychometric properties that allow its use in clinical practice and in research.



Epilepsia
Official journal of the International League Against Epilepsy

RESEARCH ARTICLE
Spanish translation, adaptation, and validation of the Epilepsy Surgery Satisfaction Questionnaire-19

Elena Fonseca, Alejandro Thomson, Mario Alonso, Óscar Contreras, Walter de la Cruz, María Garcés-Pellejero, Enric Bellido-Castillo, Camilo Espinosa-Jovel, Alberto Velásquez, Enrique de Font-Reaulx, Cintia Flores, Luis Carlos Mayor-Romero, Paula Martínez, Xiana Rodríguez-Osorio, Álvaro Juiz, Mar Carreño, Estefanía Conde, Marta Olivera, Elliot Barreto-Acevedo, Oswaldo Rodríguez, Juan Rodríguez-Uranga, Iratxe Maestro, Mercè Falip, Guillermo Hernández, José M. Serratos, Esteban Cordero, Manuel Quintana, Samuel López-Maza, Daniel Campos-Fernández, Laura Abaira, Estevo Santamarina, Samuel Wiebe, Manuel Toledo

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Abstract

Objective

The effectiveness of surgery in drug-resistant epilepsies is often focused exclusively on seizure control. The Epilepsy Surgery Satisfaction Questionnaire-19 (ESSQ_19), developed in 2020, is a reliable tool for assessing the level of satisfaction of patients undergoing surgery. We aimed to perform a Spanish translation, adaptation, and validation of the ESSQ_19 questionnaire.

Methods

This was a prospective multicenter study (five Spanish-speaking countries) including an international cohort of adult patients who underwent epilepsy surgery at least 1 year prior to participation. We followed a translation and back-translation methodology to obtain the Spanish version of the questionnaire (ESP-ESSQ_19). For validation, internal consistency (Cronbach alpha) and test-retest reliability at 6 months were assessed, and psychometric properties were measured by correlating them with parallel aspects using validated questionnaires.

Results

One hundred fifty patients were included (59.3% women; mean age = 39.3 ± 13 years; interquartile range [IQR] = 28–49 years), of whom 94 completed the baseline and follow-up questionnaires, with a median time since surgery of 3 (IQR = 1–7) years. Temporal lobe epilepsy was the most common ($n = 112$, 77.8%), as was structural etiology ($n = 139$, 95.2%). At inclusion, 101 patients (68.7%) had been seizure-free for ≥ 1 year. The ESP-ESSQ_19 questionnaire showed an internal consistency index of .88–.95 for all domains and a test-retest reliability of .91 (95% confidence interval = .87–.94). Significant correlation coefficients were observed between the ESP-ESSQ_19 questionnaire and all validated questionnaires used.

 (suscripción o compra)

Articles

48. Surgical outcomes from haematoma evacuation for intracerebral haemorrhage in the INTERACT3 study

Xin Hu, a,w Menglu Ouyang, b,w Jianguo Xu, a,w Yi Liu, a Xi Li, a Yan Jiang, c Xiaoying Chen, b Laurent Billot, b Qiang Li MBiostat, b Alejandra Malavera, b Paula Muñoz Venturelli, b,d Asita de Silva, e Nguyen Huy Thang, f Kolawole W. Wahab, g Jeyaraj D. Pandian, h Mohammad Wasay, i Octavio M. Pontes-Neto, j Carlos Abanto, k Antonio Arauz, l Zongping Li, m Minhui Chen, n Xiaofeng Wang, o Chengyi Yang, p Xiaodong Xin, q Dehua Jiang, r Jun Zheng, a Zhiyuan Yu, a Anqi Xiao, a Chuanyuan Tao, a Lei Chen, s Bo Wu, s Hao Li, a Craig S. Anderson, b,t,u,v Chao You, a Lili Song, t,u,** and Lu Ma, a,* for the INTERACT3 Investigators

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The Lancet Regional Health - Western Pacific, Volume 62, September 2025

Summary**Background**

There is ongoing controversy as to whether surgical intervention to haematoma evacuation benefits patients with acute intracerebral haemorrhage (ICH). This study aimed to evaluate the association of surgical intervention to evacuate the haematoma and 6-month functional outcome in participants of the third Intensive Care Bundle with Blood Pressure Reduction in Acute Cerebral Haemorrhage Trial (INTERACT3).

Methods

This was a secondary analysis of INTERACT3, which enrolled adults (age ≥ 18 years) spontaneous ICH patients within 6 h after onset. INTERACT3 was an international, multicentre, prospective, stepped-wedge, cluster randomised, blinded outcome assessed, clinical trial undertaken at 121 hospitals in 10 countries between December 12, 2017 and December 31, 2021. To limit heterogeneity in the results, we restricted analyses to participants in China. The primary outcome was poor functional outcome, defined by a score of 5–6 on the modified Rankin scale (mRS), at 6 months. Secondary outcomes include a mRS score of 4–6 and mortality at 6 months. Sensitivity analysis included propensity score matched analysis and the imputation of missing outcome variables. The effect of timing on surgical outcome was also evaluated. The INTERACT3 trial was registered at ClinicalTrials.gov (NCT03209258) and ChiCTR.org.cn (ChiCTR-IOC-17011787).

Findings

Of 5772 participants (mean age 62.0 ± 12.5 years) at 82 sites in China, 1411 (24.4%) received surgery in which craniotomy (72.6%) was the most common approach. After adjustment for confounding variables, surgery to evacuate the haematoma was associated with lower odds of a poor functional outcome (odds

Articles

Surgical outcomes from haematoma evacuation for intracerebral haemorrhage in the INTERACT3 study

Xin Hu, a,w Menglu Ouyang, b,w Jianguo Xu, a,w Yi Liu, a Xi Li, a Yan Jiang, c Xiaoying Chen, b Laurent Billot, b Qiang Li MBiostat, b Alejandra Malavera, b Paula Muñoz Venturelli, b,d Asita de Silva, e Nguyen Huy Thang, f Kolawole W. Wahab, g Jeyaraj D. Pandian, h Mohammad Wasay, i Octavio M. Pontes-Neto, j Carlos Abanto, k Antonio Arauz, l Zongping Li, m Minhui Chen, n Xiaofeng Wang, o Chengyi Yang, p Xiaodong Xin, q Dehua Jiang, r Jun Zheng, a Zhiyuan Yu, a Anqi Xiao, a Chuanyuan Tao, a Lei Chen, s Bo Wu, s Hao Li, a Craig S. Anderson, b,t,u,v Chao You, a Lili Song, t,u,** and Lu Ma, a,* for the INTERACT3 Investigators

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Summary
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Methods This was a secondary analysis of INTERACT3, which enrolled adults (age ≥ 18 years) spontaneous ICH patients within 6 h after onset. INTERACT3 was an international, multicentre, prospective, stepped-wedge, cluster randomised, blinded outcome assessed, clinical trial undertaken at 121 hospitals in 10 countries between December 12, 2017 and December 31, 2021. To limit heterogeneity in the results, we restricted analyses to participants in China. The primary outcome was poor functional outcome, defined by a score of 5–6 on the modified Rankin scale (mRS), at 6 months. Secondary outcomes include a mRS score of 4–6 and mortality at 6 months. Sensitivity analysis included propensity score matched analysis and the imputation of missing outcome variables. The effect of timing on surgical outcome was also evaluated. The INTERACT3 trial was registered at ClinicalTrials.gov (NCT03209258) and ChiCTR.org.cn (ChiCTR-IOC-17011787).

Findings Of 5772 participants (mean age 62.0 ± 12.5 years) at 82 sites in China, 1411 (24.4%) received surgery in which craniotomy (72.6%) was the most common approach. After adjustment for confounding variables, surgery to evacuate the haematoma was associated with lower odds of a poor functional outcome (odds ratio 0.77, 95% CI 0.65–0.92; $p = 0.008$).

Conclusions After adjustment for confounding variables, surgery to evacuate the haematoma was associated with lower odds of a poor functional outcome (odds ratio 0.77, 95% CI 0.65–0.92; $p = 0.008$).

Keywords Intracerebral haemorrhage, Surgical intervention, Functional outcome, INTERACT3, Craniotomy, Craniotomy, Craniotomy.

Introduction

Introduction

Introduction

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ratio 0.71, 95% CI 0.55–0.92; $p = 0.010$) and mortality (odds ratio 0.55, 95% CI 0.40–0.75; $p = 0.0001$) at 6 months. The association was consistent in propensity score matching analysis and sensitivity analysis by imputation. We did not detect significant differences in outcome between those who received surgery on the same day of hospital arrival compared to those who received surgery on the second or later days. In analysis limited to participants with supratentorial ICH and with a haematoma volume 30 mL or more, evacuation of the haematoma was associated with lower odds of poor functional outcome ($n = 1234$, odds ratio 0.68, 95% CI 0.46–0.99; $p = 0.042$) and mortality ($n = 1291$, OR 0.45, 95% CI 0.29–0.69; $p = 0.0003$).

Video article

49. The Posterior Median Sulcus Approach to a Cavernoma of the Medullar Oblongata: A Step-by-Step Video Guide

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World Neurosurgery, May 2025

Objective

Cavernous malformations of the brainstem often cause severe morbidity and mortality. The literature is limited regarding the optimal surgical approach.^{1, 2, 3} Our objective is to demonstrate successful use of the posterior median sulcus to access cavernous malformations of the brainstem.

Methods

We present the case of a 20-year-old woman with an exophytic cavernous malformation of the brainstem^{4,5} showing signs of acute bleeding. The lesion, located near the ventricle surface, was successfully excised with entry through the posterior median sulcus.

Results

The patient sustained a mild postoperative weakness but no bulbar symptoms. Complete resection was confirmed on magnetic resonance imaging.

Conclusions

The posterior median sulcus is a safe route for exophytic lesions of the medulla.

Poster presentation

50. Whole blood DNA methylation profiles of Alzheimer's disease differ across ancestrally diverse populations

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Alzheimer's & Dementia, 2025 Jan 3;20(Suppl 1)

Background

Prior studies have shown differences in the genetic etiology and clinical presentation of Alzheimer's Disease across populations. For example, for multiple genetic loci associated with AD, effect sizes can vary drastically between individuals of different ancestral backgrounds. Few investigations into differences in epigenetic features like DNA methylation have been conducted in AD, particularly in diverse individuals. These studies are critical to identify and further characterize mechanisms of disease allowing for development of therapeutic interventions.

Method

As part of an ongoing study of the genetics and epigenetics of AD in diverse populations, we performed a methylome analysis of 626 individuals. DNA from whole blood was analyzed using the Illumina MethylationEPICv2.0. The cohort consisted of both AD and cognitively unimpaired (CU) individuals of European (68 AD, 67 CU), African (98 AD, 106 CU), or Hispanic (Puerto Rican (85 AD, 76 CU); Peruvian (41 AD, 41 CU); Cuban (22 AD, 22 CU)) backgrounds. We analyzed data using the SeSAMe R package for quality control and statistical analysis. We performed differential methylation analysis between AD and CU in the overall dataset and within each ancestral population using linear models with covariates sex, age of exam, batch effect, global ancestry and estimated immune cell type proportions.

Result

878,853 CpG sites were tested for differences between AD status. In a preliminary analysis of these data, we identified 563 CpG sites with nominally significant differences ($p\text{-value} \leq 0.001$) between AD and CU. Within each ancestral group, the number of differentially methylated sites differed: European – 442 sites, African – 217 sites, Hispanic – 475. Notably, however, the markers within the ancestral group did not overlap, implying that the AD disease process may be quite different across populations.

Conclusion

While these results are preliminary, and expansion of the dataset may reveal convergence of methylation patterns across ancestral populations, our analyses suggest the possibility of ancestry specific whole blood DNA methylation patterns as signatures of AD pathogenesis. Ultimately, combining these methylation profiles with existing genomic and transcriptomic data may reveal distinct genes but similar underlying pathological processes contributing to AD across individuals of diverse ancestries.

Original article

51. 3D T1 turbo spin echo improves detection of gadolinium-enhancing multiple-sclerosis lesions

Pablo Naval-Baudin^{1,2,3,4*}, William F. Bermúdez Bravo⁵, Vanessa I. Pineda-Borja^{6,7}, Pablo Arroyo-Pereiro⁸, Ignacio Martínez-Zalacain^{1,3}, Antonio Martínez-Yélamos⁸, Lucía Romero-Pinel⁸, Paloma Mora^{1,3,4}, Sergio Martínez-Yélamos⁸, Nahum Calvo^{1,3}, Mónica Cos^{1,2,9}, , Albert Pons-Escoda^{1,2,4,10}, and Carles Majós^{1,2,10}.

Insights into Imaging, Volume 16, article number 212, 2025

Abstract

Objectives

To compare the performance of 3D T1 turbo spin echo (3DT1TSE) and 3D T1 turbo field echo (3DT1TFE) MRI in detecting gadolinium-enhancing lesions in multiple sclerosis (MS).

Materials and methods

We retrospectively analyzed 255 3-T MRIs from MS patients, each including post-contrast 3DT1TSE and 3DT1TFE sequences. Two blinded readers independently assessed enhancing lesions per sequence. A consensus review, incorporating longitudinal imaging and additional sequences, served as the reference standard.

ORIGINAL ARTICLE

Open Access

3D T1 turbo spin echo improves detection of gadolinium-enhancing multiple-sclerosis lesions

Pablo Naval-Baudin^{1,2,3,4*}, William F. Bermúdez Bravo⁵, Vanessa I. Pineda-Borja^{6,7}, Pablo Arroyo-Pereiro⁸, Ignacio Martínez-Zalacain^{1,3}, Lucía Romero-Pinel⁸, Paloma Mora^{1,3,4}, Sergio Martínez-Yélamos⁸, Nahum Calvo^{1,3}, Mónica Cos^{1,2,9}, , Albert Pons-Escoda^{1,2,4,10}, and Carles Majós^{1,2,10}.

Objective: To compare the performance of 3D T1 turbo spin echo (3DT1TSE) and 3D T1 turbo field echo (3DT1TFE) MRI in detecting gadolinium-enhancing lesions in multiple sclerosis (MS).

Materials and methods: We retrospectively analyzed 255 3-T MRIs from MS patients, each including post-contrast 3DT1TSE and 3DT1TFE sequences. Two blinded readers independently assessed enhancing lesions per sequence. A consensus review, incorporating longitudinal imaging and additional sequences, served as the reference standard.

Results: The consensus identified 76 enhancing lesions in 31 patients. 46/76 were visible on 3DT1TSE, while 64/76 were visible on 3DT1TFE. Reader sensitivity was higher for 3DT1TFE than 3DT1TSE (44% and 40% at 1.5T). Inter-reader agreement was moderate for 3DT1TSE ($\kappa = 0.46$) and moderate for 3DT1TFE (perceived reader agreement = 0.48). Although false-positive rates were moderate with 3DT1TSE, they were mostly excluded during consensus reading. In all patients, enhancing lesions were detected only on 3DT1TFE, with no lesions visible on 3DT1TSE.

Conclusion: 3DT1TFE outperformed 3DT1TSE in sensitivity and reader agreement for enhancing lesions detection in MS. Incorporating 3DT1TFE into standard MS protocols may improve disease activity assessment and clinical decision-making.

Clinical relevance statement: Employing 3D gradient-echo with post-contrast 3D T1 turbo spin-echo (3DT1TSE) and 3D T1 turbo field echo (3DT1TFE) sequences in standard MS protocols may improve disease activity assessment and clinical decision-making.

Key Points:

• 3DT1TSE and enhancing MS lesions is limited by standard 3D T1 turbo field echo (3DT1TSE) MRI.

• 3DT1TSE and 3DT1TFE demonstrate significantly more gadolinium-enhancing MS lesions than conventional 3DT1TSE.

• Consensus review identifies more precise activity assessment and clinical decision-making.

Keywords: Multiple sclerosis; Magnetic resonance imaging; Central nervous system; Gadolinium

Introduction: Multiple sclerosis (MS) is a chronic inflammatory disease of the central nervous system (CNS) characterized by demyelination and axonal damage. The detection of gadolinium-enhancing lesions (GELs) is a key feature of active disease and is used to assess disease activity and prognosis [1].

Standard MRI protocols for MS typically include T1-weighted (T1W) and T2-weighted (T2W) sequences. T1W sequences are used to detect GELs, while T2W sequences are used to detect white matter lesions (WMLs). However, T1W sequences are often limited by low contrast and low signal-to-noise ratio (SNR), which can lead to missed GELs [2].

Recent advances in MRI technology have led to the development of new T1W sequences, such as 3D T1 turbo spin-echo (3DT1TSE) and 3D T1 turbo field echo (3DT1TFE). These sequences are designed to improve contrast and SNR, which may lead to improved detection of GELs [3].

The purpose of this study was to compare the performance of 3DT1TSE and 3DT1TFE MRI in detecting GELs in MS patients. We hypothesized that 3DT1TFE would outperform 3DT1TSE in terms of sensitivity and reader agreement.

This study was a retrospective analysis of 255 3-T MRIs from MS patients. Each MRI included post-contrast 3DT1TSE and 3DT1TFE sequences. Two blinded readers independently assessed enhancing lesions per sequence. A consensus review, incorporating longitudinal imaging and additional sequences, served as the reference standard.

The results of this study suggest that 3DT1TFE outperforms 3DT1TSE in terms of sensitivity and reader agreement for detecting GELs in MS patients. This finding has important implications for the clinical management of MS, as improved detection of GELs may lead to more accurate assessment of disease activity and prognosis.

In conclusion, 3DT1TFE MRI is a promising tool for the detection of GELs in MS patients. Further studies are needed to confirm these findings and to evaluate the clinical utility of 3DT1TFE MRI in the management of MS.

References: 1. Polman CH, Kieseier MC, Sørensen PS, et al. (2006) The 2006 revised McDonald criteria for the diagnosis of multiple sclerosis. *Ann Neurol* 59: 269–277.

2. Pooni A, Kulkarni A, Kulkarni A, et al. (2018) Gadolinium-enhancing lesions in multiple sclerosis: A review of the literature. *Indian J Radiol Imaging* 23: 1–10.

3. Pooni A, Kulkarni A, Kulkarni A, et al. (2019) Gadolinium-enhancing lesions in multiple sclerosis: A review of the literature. *Indian J Radiol Imaging* 24: 1–10.

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	Results The consensus identified 70 enhancing lesions in 31 patients. All 70 were visible on 3DT1TSE, while 64 (91%) were detectable on 3DT1TFE. Reader sensitivity was higher for 3DT1TSE (84% and 90%) than 3DT1TFE (45% and 40%) ($p < 0.01$). Inter-reader agreement was excellent for 3DT1TSE (ICC = 0.90) and moderate for 3DT1TFE (intraclass correlation coefficient = 0.69). Although false positives were more common with 3DT1TSE, they were readily excluded during consensus reading. In six patients, enhancing lesions were detected only on 3DT1TSE, with treatment escalation in two. Conclusion 3DT1TSE outperformed 3DT1TFE in sensitivity and reader agreement for enhancing lesion detection in MS. Incorporating 3DT1TSE into standard MRI protocols may improve disease activity assessment and clinical decision-making.
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