

NEWSLETTER

GeneStroke

The Spanish Stroke Genetics Consortium

Septiembre
2013

Nº 8

Estimados compañeros

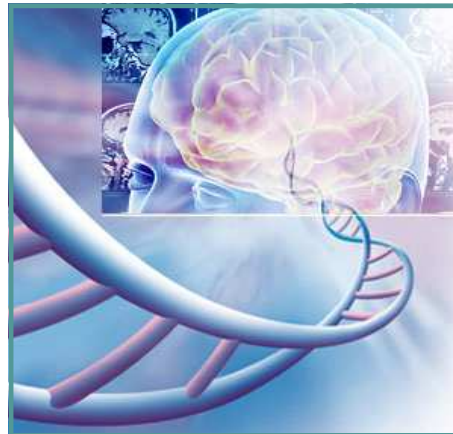
Os enviamos la Newsletter del consorcio GeneStroke, donde esperamos encontrareis información de vuestro interés, sobre las novedades del consorcio y de la genética en el ictus.

Equipo GeneStroke
www.GeneStroke.com

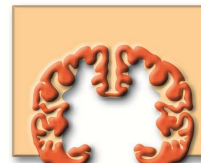
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REUNIÓN ANUAL GeneStroke



SOCIEDAD ESPAÑOLA
DE NEUROLOGIA

LXV Reunión
Anual SEN

19-23 Noviembre
2013 Barcelona

¡Os esperamos a todos!

EXTRACCIÓN DNA GRATIS !

El Consorcio pone a vuestra disposición el *Servicio de extracción de DNA* y la participación en la *Colección de muestras GeneStroke*.

Publicaciones con participación de *GeneStroke*:

Stroke Genetics Network (SiGN) Study: Design and Rationale for a Genome-Wide Association Study of IschemicStroke Subtypes.

Meschia JF, Arnett DK, Ay H, Brown RD Jr, Benavente OR, Cole JW, de Bakker PI, Dichgans M, Doheny KF, For-nage M, Grewal RP, Gwinn K, Jern C, **Conde JJ**, Johnson JA, Jood K, Laurie CC, Lee JM, Lindgren A, Markus HS, McArdle PF, McClure LA, Mitchell BD, Schmidt R, Rexrode KM, Rich SS, Rosand J, Rothwell PM, Rundek T, Sacco RL, Sharma P, Shuldiner AR, Slowik A, Wassertheil-Smoller S, Sudlow C, Thijs VN, Woo D, Worrall BB, Wu O, Kittner SJ; on behalf of the **NINDS SiGN Study**. Stroke 2013 Sep 12.

Exploring the genetic basis of stroke. Spanish stroke genetics consortium.

Giralt-Steinhauer E, Jiménez-Conde J, Soriano Tárrega C, Mola M, Rodríguez-Campello A, Cuadrado-Godia E, Ois A, Fernández-Cádenas I, Carrera C, Montaner J, Díaz Navarro RM, Vives-Bauzá C, Roquer J. Neurología 2013 June 4.

Genes involved in hemorrhagic transformations that follow recombinant t-PA treatment in stroke patients.

Fernandez-Cadenas I, Rio-Espinola AD, Domingues-Montanari S, Montaner J et al. Pharmacogenomics. 2013 April.

DNA Isolation Method is a Source of Global DNA Methylation Variability Measured with LUMA. Experimental analysis and a systematic review.

Carolina Soriano-Tárrega, Jordi Jiménez-Conde, Eva Giralt-Steinhauer, Ángel Ois, Ana Rodríguez-Campello, Elisa Cuadrado-Godia, Israel Fernández-Cadenas, Joan Montaner, Gavin Lucas, Roberto Elosua and Jaume Roquer. PLOS ONE. 2013 April 9.

PROYECTOS GENESTROKE EN ACTIVO

Actualmente tenemos estos proyectos en curso:

Proyecto: **GWALA!!** (Bases genéticas de la leucoaraiosis. Estudio de Genome Wide Association en población española)
IP: Jordi Jiménez Conde (Hospital del Mar)
Estado: En fase de imputación con 1000 genomas

Proyecto: **GWAs GenotPA**
IP: Israel Fernández Cadenas (Hospital Vall d'Hebron)
Estado: En fase de análisis

Proyecto: **GODS project** (Genetic contribution to functional Outcome and Disability after Stroke)
IP: Jordi Jiménez Conde (Hospital del Mar)
Estado: En fase de análisis de resultados de secuenciación del exoma y replicación.

Proyecto: **GLAM-Stroke** (GLObAl Methylation of ischemic stroke)
IP: Carolina Soriano (Hospital del Mar)
Estado: Terminado. En fase de publicación

Proyecto: **GRECAS Project** (Genotyping Risk and Efficacy of Clopidogrel or Aspirin following Stroke)
IP: Israel Cadenas (Hospital Vall d'Hebron)
Estado: En fase de imputación con 1000 genomas

Proyecto: **EWAS-Stroke** (Estudio de Epigenome-Wide Association en los subtipos etiológicos de ictus isquémico)
IP: Carolina Soriano (Hospital del Mar)
Estado: Financiado. Pendiente de genotipado

Proyecto: **PEPIS** (Perfil de Expresión génica y Pronóstico del Ictus iSquémico. Contribución de la expresión génica global (eQTLs) a la evolución clínica a corto y medio plazo del ictus agudo)
IP: Jordi Jiménez Conde (Hospital del Mar)
Estado: Pendiente de financiación

Proyecto: **MIXIT** (MIrna eXpression in Ischemic sTroke)
IP: Raquel Rabionet (Centre de Regulació Genòmica)
Estado: Pendiente de financiación

Proyecto: **MENEAS** (MEthylation of DNA depending on Nutrition and Exercise habits. Developing a marker of "biological Age" and risk of Stroke)
IP: Jordi Jiménez Conde (Hospital del Mar)
Estado: Pendiente de financiación

Para solicitar más información sobre los proyectos,
contactar con:



Marina Mola
(mmola@imim.es)

**¿Quieres realizar un estudio
y necesitas colaboraciones?
!!! Envía tu propuesta !!!
¡PARTICIPAD!**

NOVEDADES SOBRE GENÉTICA EN EL ICTUS:

Stroke Genetics Network (SiGN) Study: Design and Rationale for a Genome-Wide Association Study of Ischemic Stroke Subtypes.

Meschia JF, Arnett DK, Ay H, Brown RD Jr, Benavente OR, Cole JW, de Bakker PI, Dichgans M, Doheny KF, Fornage M, Grewal RP, Gwinn K, Jern C, **Conde JJ**, Johnson JA, Jood K, Laurie CC, Lee JM, Lindgren A, Markus HS, McArdle PF, McClure LA, Mitchell BD, Schmidt R, Rexrode KM, Rich SS, Rosand J, Rothwell PM, Rundek T, Sacco RL, Sharma P, Shuldiner AR, Slowik A, Wassertheil-Smoller S, Sudlow C, Thijs VN, Woo D, Worrall BB, Wu O, Kittner SJ; on behalf of the **NINDS SiGN Study**.

Stroke. 2013 Sep 12.

Abstract

BACKGROUND AND PURPOSE: Meta-analyses of extant genome-wide data illustrate the need to focus on subtypes of ischemic stroke for gene discovery. The National Institute of Neurological Disorders and Stroke SiGN (Stroke Genetics Network) contributes substantially to meta-analyses that focus on specific subtypes of stroke.

METHODS: The National Institute of Neurological Disorders and Stroke SiGN includes ischemic stroke cases from 24 genetic research centers: 13 from the United States and 11 from Europe. Investigators harmonize ischemic stroke phenotyping using the Web-based causative classification of stroke system, with data entered by trained and certified adjudicators at participating genetic research centers. Through the Center for Inherited Diseases Research, the Network plans to genotype 10 296 carefully phenotyped stroke cases using genome-wide single nucleotide polymorphism arrays and adds to these another 4253 previously genotyped cases, for a total of 14 549 cases. To maximize power for subtype analyses, the study allocates genotyping resources almost exclusively to cases. Publicly available studies provide most of the control genotypes. Center for Inherited Diseases Research-generated genotypes and corresponding phenotypes will be shared with the scientific community through the US National Center for Biotechnology Information database of Genotypes and Phenotypes, and brain MRI studies will be centrally archived.

CONCLUSIONS: The Stroke Genetics Network, with its emphasis on careful and standardized phenotyping of ischemic stroke and stroke subtypes, provides an unprecedented opportunity to uncover genetic determinants of ischemic stroke.

KEYWORDS: Cerebral infarct, genetics, genomics.

[Genome-Wide Analysis of Blood Pressure Variability and Ischemic Stroke.](#)

Yadav S, Cotlarciuc I, Munroe PB, Khan MS, Nalls MA, Bevan S, Cheng YC, Chen WM, Malik R, McCarthy NS, Holliday EG, Speed D, Hasan N, Pucek M, Rinne PE, Sever P, Stanton A, Shields DC, Maguire JM, McEvoy M, Scott RJ, Ferrucci L, Macleod MJ, Attia J, Markus HS, Sale MM, Worrall BB, Mitchell BD, Dichgans M, Sudlow C, Meschia JF, Rothwell PM, Caulfield M, Sharma P; **International Stroke Genetics Consortium.** *Stroke.* 2013 Aug 8.

Abstract

BACKGROUND AND PURPOSE: Visit-to-visit variability in blood pressure (vBP) is associated with ischemic stroke. We sought to determine whether such variability has genetic causes and whether genetic variants associated with BP variability are also associated with ischemic stroke.

METHODS: A Genome Wide Association Study (GWAS) for loci influencing BP variability was undertaken in 3802 individuals from the Anglo-Scandinavian Cardiac Outcome Trial (ASCOT) study, in which long-term visit-to-visit and within-visit BP measures were available. Because BP variability is strongly associated with ischemic stroke, we genotyped the sentinel single nucleotide polymorphism in an independent ischemic stroke population comprising 8624 cases and 12722 controls and in 3900 additional (Scandinavian) participants from the ASCOT study to replicate our findings.

RESULTS: The ASCOT discovery GWAS identified a cluster of 17 correlated single nucleotide polymorphisms within the NLGN1 gene (3q26.31) associated with BP variability. The strongest association was with rs976683 ($P=1.4 \times 10^{-8}$). Conditional analysis of rs976683 provided no evidence of additional independent associations at the locus. Analysis of rs976683 in patients with ischemic stroke found no association for overall stroke (odds ratio, 1.02; 95% CI, 0.97-1.07; $P=0.52$) or its subtypes: cardioembolic (odds ratio, 1.07; 95% CI, 0.97-1.16; $P=0.17$), large vessel disease (odds ratio, 0.98; 95% CI, 0.89-1.07; $P=0.60$), and small vessel disease (odds ratio, 1.07; 95% CI, 0.97-1.17; $P=0.19$). No evidence for association was found between rs976683 and BP variability in the additional (Scandinavian) ASCOT participants ($P=0.18$).

CONCLUSIONS: We identified a cluster of single nucleotide polymorphisms at the NLGN1 locus showing significant association with BP variability. Follow-up analyses did not support an association with risk of ischemic stroke and its subtypes.

[WellcomeTrust genome-wide association study of ischemic stroke.](#)

Markus HS. *Stroke.* 2013 June.

[Genetics of stroke: impact and limitations of evolving technology: introduction.](#)

Woo D, Meschia JF. *Stroke.* 2013 June.

CONGRESOS Y REUNIONES DE INTERÉS 2013-2014

[XXI World Congress of Neurology](#), 21-26 September, 2013. Vienna, Austria.

[Canadian Stroke Congress](#), 17-20-October, 2013. Montreal, QC. Canada.

[American Society of Human Genetics \(ASHG\)](#), October 22-26, 2013. Boston, USA.

[Society for Neuroscience \(SFN\) Annual Meeting: Neuroscience 2013](#), November 9-13, 2013. San Diego, California, USA.

[SEN 2013 - LXV Reunión Anual de la Sociedad Española de Neurología](#), Noviembre 2013. Barcelona.

[International Stroke Conference](#), February 12-14, 2014. San Diego, U.S.

[American Academy of Neurology Annual Meeting \(AAN\)](#), April 26, 2014. Philadelphia.

[23 European Stroke Conference](#), May 6-9, 2014. Nice, France.

[The European Human Genetics Conference](#), May 31-June 3, 2014. Milan, Italy.

[9th FENS Forum of Neuroscience](#), July 5-9, 2014. Milan, Italy.

[9th World Stroke Congress](#), October 22-25, 2014. Istanbul, Turkey.

GE, GE, GE...

Sugerencias...

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Estamos en la web!

www.GeneStroke.com

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