

# EAS FHSC NEWSLETTER

Issue 1, October 2016

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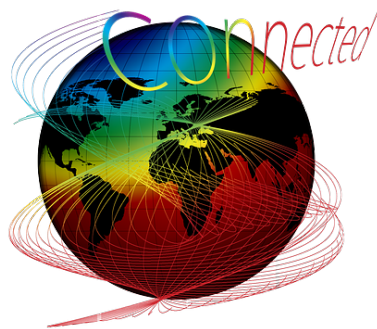
Updates from the Imperial Coordinating Centre

## EAS FHSC MISSION

**The mission of the EAS-FHSC is to empower the medical and global community to seek change in their respective countries or organizations regarding how FH is detected and managed, with a view to promoting early diagnosis and more effective treatment of this condition. Through international collaboration of stakeholders we aim to generate large scale robust data on how FH is detected, managed and the clinical consequences of current practice on outcomes.**

**Furthermore, we will work with all stakeholders including patient's organizations to ensure that these state-of-the-art information are utilized to close gaps in knowledge and/or improve clinical practice for our FH patients.**

## Message from the EAS FHSC Principal Investigator, Prof. Kausik K. Ray



About a year and a half ago Imperial College London in conjunction with the European Atherosclerosis Society (EAS) started to establish a global initiative for improving detection and treatment of individuals with Familial Hypercholesterolaemia (FH). The EAS Familial Hypercholesterolemia Studies Collaboration (EAS FHSC) is now a global network of investigators from over 50 countries worldwide who have formally committed to contributing to the initiative and this network has become the largest collaboration ever developed around FH.

The present is the 1st newsletter of EAS FHSC collaboration. We hope that this newsletter will keep you updated on the progress of the project and milestones that the consortium will achieve in upcoming months. This newsletter reflects on the many achievements of the network, and EAS FHSC most recent highlights.

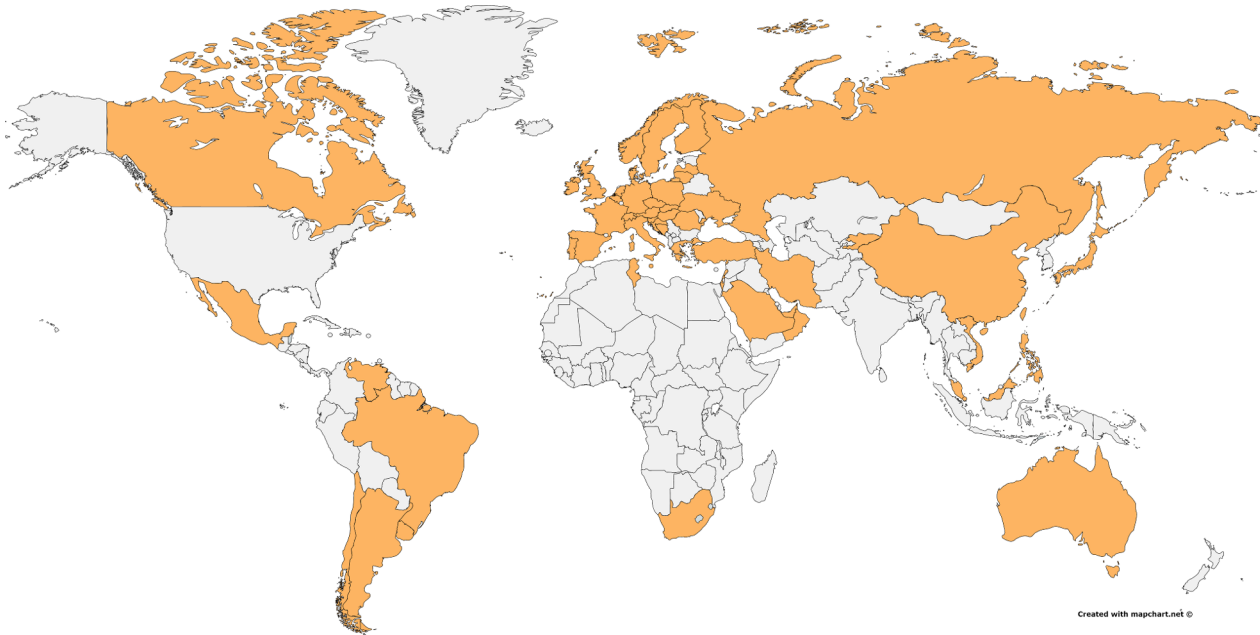
The EAS FHSC consortium, research and outreach endeavours continue to grow, and at the same time, participating

researchers continue to make significant breakthroughs in FH research. We are honoured to have a consortium of experts on FH from around the globe. The partnership among each and every lead investigator in different parts of the world is a critical component of the EAS FHSC mission. Through your support and participation, the present initiative will be able to gather data on individuals living with FH worldwide.

We have made great progress in building the bespoke secure system for data sharing and management, and we are on the cusp of implementing an application in the system for secure data sharing via logging into the website. Next year promises to be an exciting year in the progress for the project when pooling and harmonization of the first datasets received will begin. Ultimately, state-of-the-art information generated using the global data will help in promoting early diagnosis and more effective treatment of this condition.

The success of this project relies on each lead investigator and contributing investigators' dedication to the EAS FHSC mission so that we can complete the journey which we have started together. Please stay connected to us, stay informed, keep us updated and let us know what is important to you and your partners. We hope in time to highlight your work and your achievements. This is your newsletter and we are here to work with you. Thank you for your support and collaboration.

The EAS FHSC network of investigators has spread worldwide covering all the continents and this has been achieved through your continuous interest and support for the EAS FHSC collaboration.



### News from the EAS FHSC Investigators

Regular contact between the groups and the sharing of experiences between investigators and organizations from different regions and countries support the individual activities of each in driving their common objectives and help organizations consolidate their efforts and expand their scope.

**Argentina and South Africa are in the update spotlight in this issue**

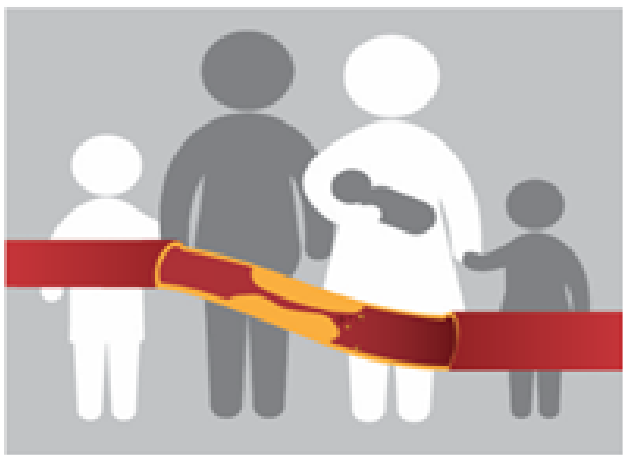
## Argentina



**By Dr. P. Corral: Argentine initiative, DA VINCI registry:** It is estimated that 82,000 to 140,000 people have FH in Argentina within a population of 41.4 million individuals. In Mar del Plata, the city where the present study was carried out, the expected number of FH patients is 1200-2000. Dr. P. Corral (FASTA University, Mar del Plata) and Dr. L. Schreier (Buenos Aires University) launched in 2015 the first FH registry in Argentina. This registry is a collaborative research between the government and a public and a private Universities: the Dept. of Health from Mar del Plata, the Laboratory of Lipid and Atherosclerosis from the University of Buenos Aires and finally, FASTA University School of Medicine.

The results were presented at the EAS Congress in Innsbruck. Among the over 51,000 adult patients included in the aforementioned database, those with a TC >300 and/or LDL-C >190 mg/dL were selected.

Almost 2000 patients met these criteria and were contacted. 226 patients were subjected to a complete screening and examination. The Dutch Lipid Clinics Criteria (DLCC) was used for FH diagnosis. Patients with a definite, probable and possible FH were 31, 24 and 105, respectively. 12 patients had secondary causes of hypercholesterolaemia. 45 patients had a first degree relative with coronary heart disease (CHD) and 32 reported a first-degree relative with elevated LDL-C levels. 6 and 26 had tendon xanthomas and premature corneal arcus, respectively. The estimated prevalence of FH in this cohort, based on definitive FH cases (DLCC), was 1 in 186. This study suggests a potentially higher prevalence of FH in Argentina than that previously estimated. The genetic studies may provide a more accurate prevalence of FH.



## South Africa

**By Prof. D. Marais:** FH is getting attention all around the world and we bring you a story from South Africa where a workshop was devoted to FH on the 24th March 2016 as part of the P5 Africa Conference and was supported by the Lipid and Atherosclerosis Society of Southern Africa.

Speakers had long been involved in research in

this field: Prof Maritha Kotze (MK), Prof David Marais (DM), Prof Derick Raal (DR) from South Africa and Prof Steve Humphries (SH) and Prof Philippa Talmud (PT) from the United Kingdom. The aim was to provide an update and perspective on FH. Though the founder effect for FH is known in the white Afrikaans-speaking South Africans, FH was subsequently identified in all sectors of the population. The methods for genetic diagnosis were reviewed by MK. The genetic investigation of subjects with a classic diagnosis of FH at a teaching hospital in Cape Town was presented (DM). The majority of the patients were white or of mixed ancestry but patients of Indian and black African ancestry also had FH. Overall about 60% of the first 1000 patients had a genetic diagnosis: LDL receptor (96%), apolipoprotein B (3%) and PCSK9 (1%). Of the 87 pathogenic mutations in the LDL receptor, the 10 commonest explained >80% of the FH and occurred in exons 4, 7, 8, and 9.

The entity of polygenic FH was discussed by SH. PM indicated the need to derive locally validated sets of genes to ensure that the diagnosis would be made appropriately in South Africa. DR reviewed the treatment of FH and indicated that most patients could, with modern treatment, achieve LDL concentrations that would dramatically lower the risk of coronary disease. More detailed information about the meeting will be published in due course. It is hoped that the research in FH can be translated to clinical practice with better awareness and treatment.



The Hellenic Atherosclerosis Society has developed a free FH diagnosis calculator tool that can be downloaded following these links

- App Store: <https://appsto.re/gr/wF4Q7.i>.
- Google Store: <https://play.google.com/store/apps/details?id=com.ajjumax.helleniccalculat>.
- Desktop: <http://web.alphabit.gr/FHCalculator/index.html>.

### PUBLICATIONS

Let us know about your recent publications and we will include the references and links in this section in the next issues of the EAS FHSC newsletter, as a means to share research and results around FH within the EAS FHSC network.

### EVENTS

#### **FH Awareness Week**

Hope every one celebrated FH-awareness day on September 24, 2016 and awareness week (19-25 Sept). Please click on the link to find further details of events. The EAS produced and distributed a daily newsletter and placed a list of national events carried out by different investigators worldwide.

[http://www.eas-society.org/?page=fh\\_week\\_2016](http://www.eas-society.org/?page=fh_week_2016).

#### **Educational Programme on FH**

A 2-day EAS advanced course on FH will take place in October 13-15, 2016, in Athens, Greece.

[http://www.eas-society.org/page/course\\_FHAthens](http://www.eas-society.org/page/course_FHAthens).

#### **EAS FHSC Central and Eastern European Symposium on FH**

The EAS FHSC Central and Eastern European Symposium on FH will take place between 25-27 November, 2016 in Warsaw, Poland. [http://www.eas-society.org/page/Symposium\\_fh\\_welcome](http://www.eas-society.org/page/Symposium_fh_welcome).

#### **85th EAS Congress**

The 85th EAS Congress will be held in Prague, Czech Republic, between 23-26 April, 2017.

<http://eas2017.com/>.

#### **Contact**

Let us know about your news, publications, achievements or events around FH and we will include them in the following issues of the EAS FHSC newsletter, so they can be shared with the rest of colleagues, given publicity and contribute to networking within the EAS FHSC collaboration.

[info@eas-fhsc.org](mailto:info@eas-fhsc.org).

[coordinator@eas-fhsc.org](mailto:coordinator@eas-fhsc.org).



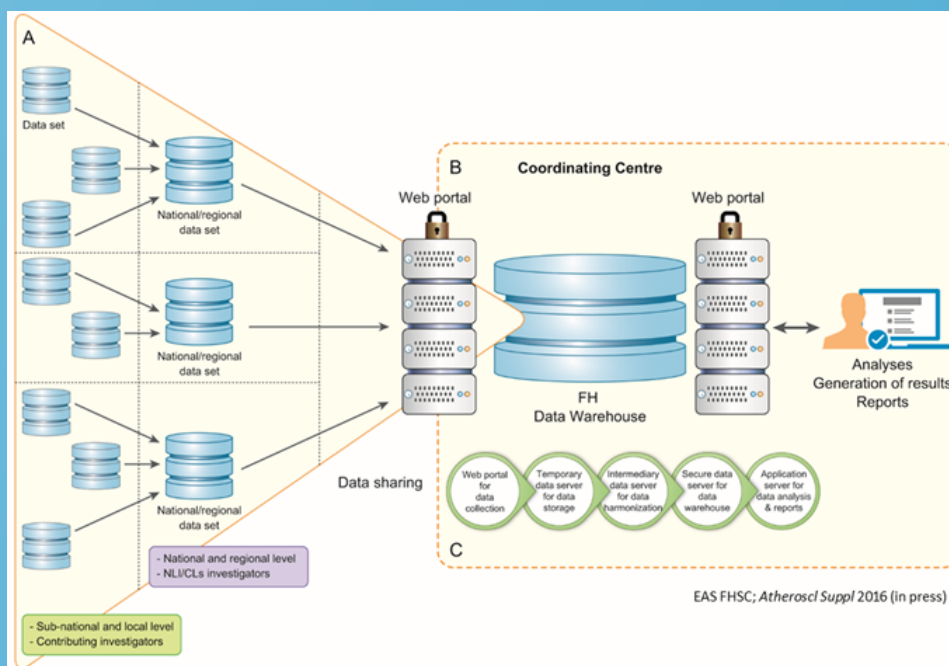


The rationale and methods paper for the EAS FHSC collaboration was recently accepted for publication at *Atherosclerosis* and will be released in the upcoming weeks. This paper also includes the protocol for the EAS FHSC project. We are very grateful to all the EAS FHSC lead investigators for your contribution, comments and critical review of this important paper for the collaboration.

The team at the Coordinating Centre is at your disposal in assisting with queries and facilitating data management and sharing. We intend to work with all the investigators involved in the EAS FHSC initiative to help in the contribution to the EAS FHSC initiative.

## BIOINFORMATICS UPDATE

The IT team at the Coordinating Centre is building a central bespoke data warehouse that will be able to consolidate data from multiple sources and formats into a robust, large scale FH registry. The system will provide through novel informatics the capacity for retrospective and ongoing prospective data pooling, harmonisation, and analysis. As a first step, we will soon be able to share a secure data sharing application via which datasets from partners can be securely uploaded to the FH Data Warehouse at the Coordinating Centre, ICL. The details of the system are included in the EAS FHSC protocol that will be published in the journal *Atherosclerosis* in the following weeks.



Thank you very much!