

EAS FHSC NEWSLETTER

Issue 10, November 2019

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FHSC Data Entry Application – Version II

FHSC Data Entry Application Version One, released last year, met its expectations and Version Two is currently being developed to bring about improvements including:

- Ability for centres within multicentre registries to collaborate and enter patients in parallel.
- Reduce data entry burden by deriving fields automatically whenever possible (e.g. BMI derived from height and weight).

It is anticipated and strongly encouraged that all new data submissions to the FHSC Global Registry will be via the use of this Application to facilitate uniform, standardised data formats.

EAS FHSC spans 69 countries as shown in the shaded map below, includes 87 Lead Investigators which are listed [here](#), and the Global Registry now has **61,650 cases** recorded across 59 countries



More about the EAS FHSC is available via the [website](#).

FH Studies Collaboration

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Follow us at:

[#fhscglobalregistry](#)



www.eas-society.org/fhsc

The FHSC was represented by the Coordinating Centre Investigators from Imperial College London UK, on behalf of FHSC Investigators

ESC Congress/World Congress of Cardiology, Paris, France 2019

Dr. Antonio J. Vallejo-Vaz

FHSC Clinical Research Fellow, gave a talk entitled:

'The EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC) global registry: Analyses from over 55 000 cases and 68 countries'

ESC = European Society of Cardiology

ESC Digital Summit, Tallinn, Estonia 2019

Christophe Stevens

FHSC Data Manager and Software Developer, gave a talk entitled:

'Novel bespoke IT solutions to harmonise and pool heterogeneous datasets and cohorts from multiple countries into a single registry: The Harmonised Application and Registry of Patients (HARP)'

FH Europe Annual Network Meeting, Bucharest, Romania 2019

Dr. Alexander Lyons

FHSC Research Associate Project Manager, gave a talk entitled:

Keynote II: 'EAS FHSC Global Registry'

Become part of the EAS FHSC

Are you a researcher with an interest in FH, collect clinical and/or genetic FH data and keen to contribute to the EAS FHSC Global Registry?

If so, more information can be found in the open-access publication of the FHSC Study Protocol ([click here](#)) and the EAS FHSC Coordinating Centre would like to hear from you.

For enquires and discussions, contact the FHSC Coordinating Centre via info@eas-fhsc.org

What's new in FH in Canada?



The FH Canada registry created a network of academic clinics across Canada with the common objective to improve the identification of patients with FH and initiate personalized treatment. The national registry now includes over 4,400 patients. A new simplified and practical definition for FH, with diagnostic performance comparable to existing criteria but adapted to the Canadian population, was recently validated and published in the Canadian Journal of Cardiology (Figure 1). To make it easier to use with patients, the Canadian definition has been incorporated into the Ca-

-rdioRisk App, under "FH Calculator", which also includes an algorithm to impute a baseline LDL-C from an LDL-C while on treatment when the original baseline LDL-C can't be retrieved. The CardioRisk App is freely available to all health care professionals; it generates a report to be saved and added to patient's file. It is available for computers at <http://www.circl.ubc.ca/> or for smartphones in App Store or Google Play. These recently developed tools facilitate the clinical diagnosis for FH and help clinicians to initiate cascade screening.

By Dr. Jacques Genest, NLI Canada

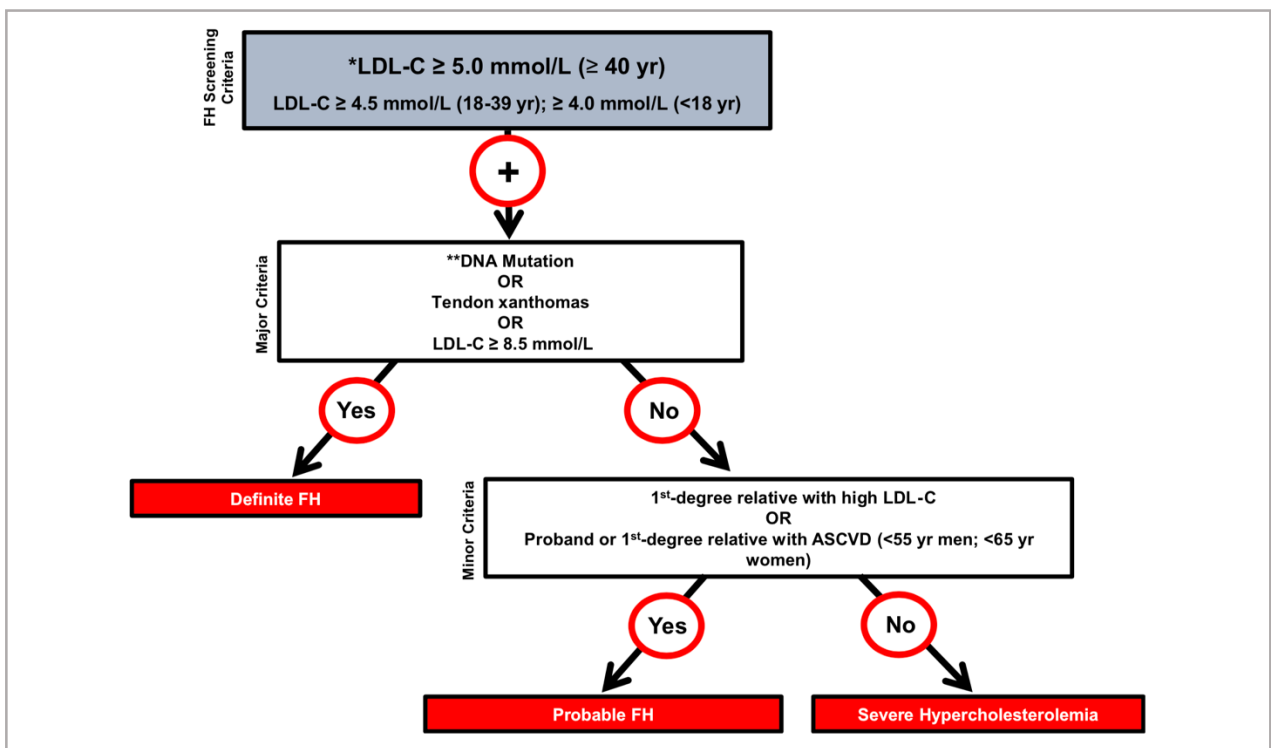


Figure 1. Canadian definition for the clinical diagnosis of familial hypercholesterolemia

* Secondary causes of high LDL-C should be ruled out (severe or untreated hypothyroidism, nephrotic syndrome, hepatic disease [biliary cirrhosis], and medication, especially antiretroviral agents).

** Causal DNA mutation refers to the presence of a known FH-causing variant in the LDLR, APOB, or PCSK9 gene in the proband or a first-degree relative.

Reproduced from Ruel *et al. Can J Cardiol.* 2018;34(9):1210-1214 with permission from Elsevier.

Isabelle Ruel, Liam R. Brunham, G.B. John Mancini, Robert A. Hegele, and Jacques Genest, on behalf of FH Canada investigators.

Vroni: Early Screening for FH



Heterozygous FH represents the most frequent genetic disorder with an estimated prevalence of 1:250 in the general population. Early diagnosis enables the initiation of preventive measures already in childhood leading to the prevention of severe consecutive atherosclerotic manifestations. In Germany, as in most European countries, there is no early detection programme for FH in place.

As part of a digital medicine programme, DigiMed Bayern, the Vroni study is sponsored by the Bavarian Ministry of Health, and will offer all children living in the Free State of Bavaria a screening for FH in the context of obligatory paediatric visits (U9 – J1) in the age group of 5-12 years. 200 µl of capillary blood will be taken with a finger prick blood test and LDL-C will be measured by a standard automated method. In case of a value above the 95 percentile of the age specific distribution, molecular genetic analysis for FH will follow. Children tested positively and their parents will be informed comprehensively about the

disease at the time the diagnosis is established and they will be transferred for further care to the regional paediatric cardiologist and to specialized training centres.

Taking the index patient (child) as the basis we plan to establish a reverse cascade screening programme in first degree relatives and further relatives to identify additional subjects with FH. Long-term aim of the study is an improvement in the diagnosis and treatment of patients with FH in Germany. The Vroni study will also address in detail the epidemiology of FH in Germany.

By Prof. Dr. Heri Schunkert, NLI Germany



V.Sanin, C.von Hagen, G.Leipold, K.Appel, V.Mall, Friedmann, F.Wippermann, H.Schunkert

Egypt: EAVA Society



The Egyptian Association of Vascular Biology and Atherosclerosis (EAVA) led by Prof. Ashraf Reda has conducted the largest cross-sectional study across Egypt to evaluate the risk factors' profile among Egyptians presenting with acute coronary syndromes (ACS). From November 2015 to August 2018, data were collected from 3,224 patients with ACS in 30 coronary care centres covering 11 governorates across Egypt.

The investigators presented a poster abstract in the last 2019 ESC congress held in Paris, in which they showed a unique dataset focusing on premature coronary artery disease (CAD). Remarkably, the prevalence of premature CAD in Egypt (defi-

ned as ACS before age 55 years in males and 65 years in females) is high (51%). Risk factors' (RFs) distribution showed a significantly higher prevalence of Diabetes Mellitus, obesity, and smoking in patients with premature CAD when compared to others. Interestingly, rates of documented dyslipidemia were similar between groups with no significant difference in low-density lipoprotein-cholesterol levels.

EAVA is also working with the EAS FHSC to generate robust information to accurately and reliably investigate the burden of FH, and the clinical consequences of current practices on delivery of care.

By Prof. Ashraf Reda, NLI Egypt



Official Iraqi channel contributes to FH awareness



Since the start of 2018, an extensive FH awareness programme was launched in Iraq. Many community layers were included (specialist, GPs, teachers of primary schools, university staff, engineers, lawyers and even army soldiers). These great efforts succeeded in sharing knowledge and raising awareness about FH and its public health burden on the Iraqi community.

Recently, our voice reached the Iraqi Ministry of Health, which invited us to talk about this issue and to suggest strategies to tackle FH in Iraq.

On 24th September, the official Iraqi channel 'Iraqia TV' invited us, for the first time, to talk to the Iraqi people via this channel about FH including its prevalence, impact, importance of early diagnosis and proper treatment as well as our strategies to decrease its burden in the country.

Fifteen minutes of TV airtime allowed for this lay talk, which was supported by images of skin manifestations of the disease. This interview was posted on many social media platforms to be accessible to the public.

By Dr. Mutaz Al-khnifsawi, NLI Iraq



Please follow the link to watch the interview <https://youtu.be/fe2P8h7PELE>

FHSC Steering Committee

**FOR THE ATTENTION OF
The FHSC Steering Committee Members**

The EAS FHSC Steering Committee annual closed meeting of 2020 will take place in conjunction with the EAS 2020 Congress (31st May – 3rd June) in Geneva, Switzerland
on

Sunday 31st May 2020, from 09:00 to 14:00 hrs

This closed meeting is facilitated by the EAS FHSC Coordinating Centre and welcomes, **by strict invitation only**, EAS FHSC Steering Committee members and invited participants.

The agenda includes updates on the FH registry and associated projects from the FHSC Coordinating Centre of Imperial College London, UK and a series of presentations from invited speakers yet to be confirmed.

