LETTER TO THE EDITOR

Dutch national screening and disease management program for Familial Hypercholesterolemia (FH) – A model for Saudi Arabia?

Genetic Screening for and Disease Management of FH is an underserved area in need of more attention in the Kingdom and Gulf Region. A well organized, comprehensive disease management program initiated early in the disease process can substantially improve outcomes. Such a program is also essential in secondary prevention for FH patients who require surgical intervention.

In the Netherlands a cascade genetic screening program to trace family members with Familial Hypercholesterolemia has been successfully implemented by the StOEH (Stichting Opsporing Erfelijke Hypercholesterolemie) since 1994. Over 45,000 Dutch FH family members have been tested and approx. 18,000 have been diagnosed using DNA diagnostics. The strategy of using cascade family screening and DNA analysis as a diagnostic tool is very cost effective as it minimizes expensive hospital visits. Patients contacted appreciated this direct approach which was reflected by a >90% participation rate and a sharp increase in the number of patients being treated with cholesterol lowering drugs from 39% at the screening visit to 93% after a one year evaluation. A recent 10 year follow up of diagnosed FH patients showed a dramatic improvement of their survival that was comparable to age and sex matched non-affected controls in the Rotterdam study.

Patients with confirmed diagnosis are followed in focused disease management program that addresses the correctable lifestyle issues and provides optimal medical therapy. Those that fail these measures or confirmed homozygous FH patients are considered for advanced therapeutic options like LDL-apheresis. For Homozygous FH patients, except for combined heart–liver transplant, this is the only available treatment to prevent cardiovascular death in adolescence or early youth.

In the Netherlands, with 16.5 million inhabitants, the screening program has been successfully executed as a national screening program for the last 6 years with a relatively small staff of 12 FTE local genetic field workers and 2.5 FTE centrally located support staff.

The StOEH program is considered a successful strategy that is now being replicated in the UK, and Norway, France, Austria, Belgium. Taiwan has also shown interest to emulate the Dutch approach as well.

Given that the majority of the complications of this disorder are premature cardiovascular events and the national scope of the problem the Saudi Heart Association is the logical forum for such a program.

It is important to note that high tech and medical solutions in the treatment of FH while important, should not be the initial focus. The starting point of a program should focus on education of families regarding diet, exercise and other modifiable lifestyle factors. Having a trained FH disease management expert visit the family in their homes is very important and allows national coverage. All interventions must be evaluated to obtain clear evidence of safety, efficacy and cost effectiveness. Using a centralized data registry for cascade screening of family members as well as monitoring diagnostic and therapeutic interventions would improve efficacy and quality of individualized treatment. In the future this would enable in depth analysis of the effectiveness of the program as well as the management of individual cases.

Using a proven model for FH disease management that follows treats and refers patients allows for improved FH outcomes and more rational utilization of tertiary care facilities and resources. Such a program is achievable with national collaboration. The expertise and experience gained from the FH screening and disease management program could serve as a model for other orphan diseases and larger disease management initiatives for which good treatment options are available.

Integrated disease management program should be a starting point for treating patients with FH. Advanced options for management should then be carefully planned on a regional basis to avoid duplication of services and optimize return
(improved outcomes) on human and capital investments. All interventions must be monitored to improve and optimize outcomes. Based on this model, regional funding would be allocated to support services like apheresis in dedicated facilities where expertise of the professions, engineers and others would provide the critical mass staff needed to provide a quality service. Such regional services would likely be much more cost effective.

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Joseph G. Franke
Cardiac Clinical and Research Information Systems Coordinator,
Cardiac Sciences,
King Abdulaziz Cardiac Center,
King Abdulaziz Medical City,
Riyadh,
Saudi Arabia.
Tel.: +966 12520088x16739 (office);
mobile: +966 502315291.
E-mail addresses: frankej@ngha.med.sa, frankej@gmail.com

Peter J. Lansberg
Coordinator Durrer Center for Cardiogenetic Research,
Dept. of Vascular Medicine,
Academic Medical Center – Amsterdam,
Meibergdreef 9, F4-159.2,
P.O. Box 22660,
1100 DD Amsterdam,
The Netherlands.
Tel.: +31 20 566 2239/6612, +31 654 347 391 (mobile);
fax: +31 20 566 9343
E-mail addresses: p.j.lansberg@amc.uva.nl,
lansberg@gmail.com
Available online 31 October 2009