

COST ACTION BM083

“A European Network of the HLA Diversity for Histocompatibility, Clinical Transplantation, Epidemiology and Population Genetics (HLA-NET)”

Meeting title: Anthropological aspects of BMD registries
Reference: ECOST-MEETING-BM0803-301012-023056
Meeting dates: from 30 to 31 October 2012
Organisation: Dr Constantia Voniatis Papioannou
Location: Makarios III Hospital, Nicosia, Cyprus

SCIENTIFIC REPORT

A two-day symposium called *Anthropological aspects of BMD registries* was organized in Cyprus on 30-31 October 2012 in order to address and discuss several methodological issues raised by the complexity and heterogeneity of the HLA data stored in European Bone Marrow Donor (BMD) registries. Indeed, many statistical and bioinformatics tools are currently available to analyze complex HLA data including typing ambiguities and a main question is to know whether such tools can be applied to registry data in the same way they are applied to well-defined population samples, e.g. can we deal with the high quantity of ambiguities and mixed resolution levels characterizing these BMD data sets to estimate gene frequencies and other parameters? Are BMD data representative of local populations? Can we use these data in research studies aiming at comparing the HLA genetic diversity among different populations? Also, a question of great interest was to understand the current situation in terms of BMD registries management, funding, data availability for transplantation, etc, in different European countries.

On the first day, the meeting started with presentations on several topics related to BMD registries, genetic diseases and HLA diversity in Cyprus by colleagues working in diverse institutions of the island. Dr Constantia Voniatis Papaioannou (Medical and Public Health Services) presented the Cyprus experience in terms of histocompatibility, including historical aspects, methodologies used, benefits in transplantation and diseases, and current activities; Professor Constantinos Deltas (Laboratory of Molecular and Medical

Genetics, University of Cyprus) made a state of the art on genetic diseases in Greek Cypriots and the diversity of their gene pool; Dr Paul Costeas (Karaiskakio Foundation) explained the situation of the BMD Cyprus registries with a historical perspective; and Dr Agathi Varnavidou (Histocompatibility and Immunogenetics Laboratory, Nicosia General Hospital) talked about HLA-B27 and disease associations in Greek Cypriots. This gave all participants a thorough idea of Cyprus participation to several domains related to the subject of the symposium.

In the afternoon, the presentations focused on data analyses recently performed on several BMD registries in Europe: the Swiss registry, with the main methodological part presented by Dr José Manuel Nunes and the results of data analyses by Dr Stéphane Buhler (University of Geneva); and results on the Slovenian registry presented by Dr Blanka Vidan-Jeras (Blood Transfusion Center of Slovenia). In addition, a study on the registry from Quebec was presented by Dr Lucie Richard (Héma-Québec) who also described the history of colonization of Quebec by French settlers during the 16th and 17th centuries. These talks showed that BMD registry data can be used successfully to investigate the HLA genetic diversity patterns of different countries, with an anthropological (human peopling history) perspective.

The second day started with the presentations of two other registries whose data analyses are currently starting: the French Rhône-Alpes registry presented by Dr Valérie Dubois (EFS, Lyon) and Dr Stéphane Buhler (University of Geneva); and the Belgian registry presented by Dr Michel Toungouz Nevessignsky (ULB, Brussels). In addition to anthropological and biostatistical aspects, these presentations gave rise to discussions on other, more societal/political topics such as the status of small versus large registries in Europe, the problems of donor recruitment, the institutional and/or governmental supports, etc. Finally, thanks to a teleconference with Dr David Townend (Maastricht University), ethical aspects related to the use of BMD registry data were discussed, e.g. questions and problems about informed or broad consent of sampled individuals, benefit sharing, commercial use of information, etc.

Overall, this symposium was highly informative as it addressed very diverse topics related to BMD registries in Europe and stimulated the participants to collaborate and use both consensual approaches and standard protocols for data analysis, as those proposed by HLA-NET.