2nd Conference of 'EB-CLINET – Clinical Network of EB Centres and Experts'
September 17 – 18, 2013 – Salzburg, Austria

EB-CLINET CONFERENCE REPORT

The second annual meeting of EB-CLINET was held at the General Hospital (Paracelsus Medical University) Salzburg from 17th-18th September 2013, attracting 91 delegates from 29 countries across the world. The meeting was funded by DEBRA Austria with additional sponsorship from Interspar, Flen Pharma and PolyMem. CAP Partner supported the conference organisationally.

The conference was opened by Dr Stöckl (Member of the County Government of Salzburg) who welcomed delegates to the hospital. He congratulated the EB House Austria on its excellent clinical, research and educational activities, and expressed his pleasure that Salzburg was the venue for this important meeting. Dr Rainer Riedl (CEO of DEBRA Austria, Managing Director of the EB House and President of DEBRA International) welcomed delegates on behalf of DEBRA Austria/ EB House Austria and DEBRA International, noting that EB-CLINET complemented the long-standing patient and research networks so that all stakeholders in EB have an opportunity to collaborate.

Prof. Helmut Hintner (Head of the Department of Dermatology, General Hospital, Paracelsus Medical University Salzburg) then gave his keynote speech, from the perspective of a leading EB clinician and as a member of the European Union Committee of Experts on Rare Diseases (EUCERD), on European Reference Networks (ERNs) in the context of rare diseases. He emphasised the opportunity that EB-CLINET has provided a model for the foundation of an ERN for Genodermatoses (GERN). He reviewed the essential elements of a successful ERN, which include a substantial patient base, multidisciplinary care, basic and clinical research, patient group involvement, publications and a comprehensive communication network. He believed that the proposed GERN meets these criteria and is a good candidate to be one of the first ERNs to receive EU funding.

Dr Gabriela Pohla-Gubo (Executive Manager of the Academy at the EB House Austria and Project Leader of EB-CLINET) gave a brief overview of the activities of EB-CLINET since the inaugural meeting in 2012. An impressive international coverage, with 52 member institutions from 43 countries, has been achieved and several work packages, such as the development and elaboration of registries, are already in progress. Others, such as creating directories of centres of expertise and specialist laboratories, will commence over the coming months. Workshops and training courses have also been held under the EB-CLINET umbrella. A questionnaire updating currently held information on the member groups had recently been circulated and she encouraged all member institutions to respond.

Picking up on the comments on registries, Dr Peter van den Akker (Department of Genetics, University Medical Centre Groningen, The Netherlands) gave an update on the work stream on registries which he coordinates on behalf of EB-CLINET and, in particular, on the outcome of a workshop held in April 2013. This workshop had considered various strategies for taking the project forward and had

recommended that dystrophic EB (DEB) be the focus of the first efforts, building on the DEB mutation database pioneered in Groningen and his existing collaboration with the Polish Institute of Mother and Child. It is proposed that a three year project be established to create an overarching framework to connect locally held databases and define a minimum data set. He acknowledged that there are many challenges ahead, such as funding and data protection regulations, but he believed that much valuable data could be gathered, including patient-reported data from EBCare. Dr Katarzyna Wertheim-Tysarowska (Department of Medical Genetics, Institute of Mother and Child, Poland)

elaborated on the proposal by detailing her experience in establishing a mutation-specific database for DEB, emphasising the need for international collaboration and data sharing.

Delegates then divided into smaller workshop groups to explore in detail what the minimum data set should comprise, the features needed by clinicians, researchers and patients from the proposed register and the contribution that each delegate and their institution could make to its development. This short summary does not allow a full report to be given on each workshop but more information can be obtained from the conference organisers.

Emphasising the essential link between research and clinical practice, Dr Clare Robinson (UK), the DEBRA International Research Manager, gave DEBRA's perspective on driving research outcomes through to clinical practice. After giving an overview of the many potential therapies in late preclinical or early human trial, she described DEBRA's commitment to translating the most promising of these into the clinic. She noted that basic and clinical research is still needed in respect of all of the current candidate therapies but also stressed that the patient group is increasingly focused on how to mobilise funding and expertise to take therapies to market. She made reference to the increasingly important use of advisers from industry, venture capital and regulatory agencies and expressed the patient groups' pleasure in the increasing interest of biopharma in EB.

She was followed by Dr Lawrence Charnas (USA) of Shire Pharmaceuticals who spoke about his company's programme to develop protein therapy for DEB. After describing the regulatory pathways, he stressed the importance of natural history studies to help define clinically meaningful end points for clinical trials. He also outlined some of the scientific and regulatory challenges that lie ahead before a therapy can be licensed and reimbursed.

A further live case study was given by Dr Paul Kemp (UK) of Intercytex Ltd on his collaboration with Prof. John McGrath in London, following EB2006, using injectable allogeneic human dermal fibroblasts in the treatment of DEB. The Phase II trial has now been completed and the results show that there is an improvement in wound healing for the first 28 days. The product is now available in the UK as an Article 5 'special' and he hoped that clinicians in other countries would use this route to treat patients and increase the available data since the therapy is still evolving. He noted that recruiting sufficient patients had been a challenge and that the number of wounds studied had been below the original project protocol as a result. His current plans now include extending the shelf life of the fibroblasts and developing a pain-free method of delivery.

The second day commenced with a keynote speech from Dr Susanne Krämer (Faculty of Dentistry of the University of Chile) on the importance of clinical practice guidelines, which are being developed by DEBRA International, to ensure that clinical care is guided by evidence-based practice and by expert opinion. She described the challenging and laborious process that has to be undertaken to provide such a guideline.

Dr Anja Diem (EB House Austria) gave a presentation on the patient handbook which has been written at the EB House. This handbook seeks to provide information for patients on managing EB and living with EB and complements the clinical practice guidelines aimed at professional audiences. Dissemination will primarily be by a dedicated website and the handbook is currently available in German and English. Translation into other languages will be welcomed if funding can be found.

Prof. Leena Bruckner-Tuderman (Head of the Department of Dermatology, University Medical Centre Freiburg, Germany) then spoke about EB-CLINET's plans to increase opportunities for professional training. The modalities being considered include courses, hands on visits of observation and, eventually, visits by a group of EB specialists to countries wishing to learn on their home ground. It was felt that the larger EB Centres of Expertise would be the most appropriate to initiate training in the first instance.

The sessions on clinical practise guidelines and professional´s training were followed by workshops to develop ideas on future content and methodology. Again, space does not permit fuller description of the outcomes which are available from the organisers.

In addition to the presentations on the topics described above, a series of clinical case presentations and case studies were made and discussed throughout both days. Presenters from Austria, Belgium, Czech Republic, Chile, Egypt, Germany, Ireland, Italy and the Ukraine led these sessions and their slides, along with other presentations are available on the EB-CLINET website.

The next EB-CLINET conference will be held in London in September 2015.

For further information please contact Gabriela Pohla-Gubo (**g.pohla-gubo@salk.at**) or visit the EB-CLINET website (**www.eb-clinet.org**).

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