

Gaucher Masterclass in South Africa

Tanya Collin-Histed reports:

On Tuesday 17th October, Tanya Colin-Histed of the EGA, Prof Chris Hendriksz, Dr Greg Pastores and Dr Hylton Sevitz ran a MASTERCLASS in Cape Town South Africa. This was joint venture between the EGA and the UK Gauchers Association who provided funding through the Susan Lewis Memorial Fund to support many of the attending doctors.

The Gaucher Masterclass attracted physicians from all over Africa including Cameroon, Botswana, Sudan, Rwanda, Malawi, South Africa and Zimbabwe. Rare Disease Botswana were also represented by their cofounder and mother of two children with a rare disease and Aimee-Kate Bosch, a Gaucher patient and a member of the Gaucher and LSD Society of South Africa.

The idea for this masterclass came from a) the EGA's work in educating doctors and b) following several applications to the EGA from several different countries in Africa for help in getting access to ERT through charitable access programmes as Governments in these countries do not provide the treatment through public health programmes, although a few patients in South Africa are now receiving ERT from the Government, these are in the minority. As these doctors are only treating one or two patients it is important to a) improve their knowledge of the disease for diagnosis and clinical management and b) to develop a sustainable network for ongoing support and guidance. Prof Chris Hendriksz led the masterclass and donated his time to develop the programme and identify the speakers. Chris, a doctor from one of the LSD treatment centers in the UK, South African born and his wife Flo have set up a company that are making educational videos to support the development of knowledge and clinical practice guidance for metabolic conditions and they videoed the masterclass to provide ongoing tutorials for the participants that they can tap back into.

The day focused on areas such as an overview of the disease and its pathology, highlighting its clinical signs and symptoms, a description of the different subgroups of the disease and the challenges of diagnosis. One of the morning sessions led by Dr Karen Fieggen highlighted the challenges of diagnosis in an environment where many children and adults had other clinical/health challenges such as TB, HIV and malnutrition. Topics such as co morbidities, the use of biomarkers and different types of therapeutic options were discussed, often with lots of questions during the sessions which stimulated a lot of very valuable input and awareness of local challenges.

In the afternoon, the focus was on supporting the patients and families, the development and sharing of clinical guidelines and the importance of building networks in countries. I finished the day with a presentation of the value of working with the patient organization outlining my story as a mother, which is often at the heart of many rare disease patient groups, the work of the UK Gauchers Association as an example of how to support patients and their families and then the strategic role of the EGA.

The Feedback from the masterclass has been very positive, both on the day and through the feedback forms we have received back from the participants.