The International Gaucher Alliance (IGA) is an umbrella organization representing 56 patient associations from all over the world. The regional manager (RM) program was developed to provide additional on the ground resources and are the eyes and ears of the IGA in the many different countries that have Gaucher patients but do not have any formal patient organization or where there are many challenges.

**CASE STUDY: NEPAL**

At the beginning of November 2019, IGA Regional Managers working with Dr Ashok Vellodi, a retired paediatric consultant from Great Ormond Street Hospital, London (working as a volunteer) visited Kathmandu, Dharan and Biratnagar in Nepal.

**LEARNING POINTS & NEEDS**

- Getting the DBS kits and sending samples back to neighbouring country India is very challenging
- Changing the attitude of doctors in Nepal to these rare disorders
  - Lack of genetic training among doctors
  - Lack of awareness among doctors
  - Lack of infrastructure and proper framework for diagnosis of LSDs
  - Lack of awareness and knowledge among parents/caregivers
  - Need for patient support groups
  - Need for better coordination and coherence among doctors from different cities
  - Need for humanitarian aid to diagnosed patients

**TARGET OUTCOMES:**

During the visit, Dr Vellodi instructed the residents on history and examination techniques and provided management advice, followed by a meeting about the challenges facing doctors in Nepal. Patients and families with diagnosed lysosomal storage disorders (LSDs) came to meet us and allowed Dr Vellodi to examine the patients. Over 50 paediatricians from hospitals in Kathmandu and over 30 from Biratnagar hospitals attended the program which included lectures from Dr Vellodi covering case-based approaches (including a live case-based approach on two Niemann-Pick siblings), accurate diagnosing and better management of LSDs.

**PROBLEMS HIGHLIGHTED BY FAMILIES:**

- No access to treatment
- Follow up testing is a huge challenge
- Cost is a huge challenge in managing a disease

www.gaucheralliance.org

**THEME 7: RARE DISEASE PATIENT GROUPS INNOVATIONS POSTER 274**

**AUTHORS**

T Collin-Histed1 International Gaucher Alliance (IGA), Gloucestershire, UK
V Aleksovska2 International Gaucher Alliance (IGA), Gloucestershire, UK
S Sathe3 International Gaucher Alliance (IGA), Gloucestershire, UK
S Tyagi4 International Gaucher Alliance (IGA), Gloucestershire, UK