

Sema Berktas, MD.  
Paediatric gastroenterologist and hepatologist  
Turkey

ESPGHAN Head Office  
Durford Mill,  
Petersfield, Hampshire  
GU31 5AZ, United Kingdom

Dear ESPGHAN committee members,

I have spent three intense months from 4th April to 4th July 2016 at the Translational Gastroenterology Unit & Department of Pediatric Gastroenterology and Nutrition, the University of Oxford Hospitals NHS Trust, Oxford, United Kingdom under the guidance of Professor Holm Uhlig combining research and learning paediatric gastroenterology.

During my stay, I participated in a research project between Holm Uhlig and international collaborators which focused on a new genetic defect in IL11 signaling that presents with severe immune dysregulation including extreme food allergy and immunodeficiency with viral immune problems. We collected phenotype and functional data from patients who consented as part of the research project. IL11 signalling has recently attracted attention since there is dominant signature of IL11 signalling in patients with IBD (West et al. Nat. Med 2017). We therefore investigated patients with IL11RA defects who have a loss of function defect. In one patient a severe Th2 mediated inflammation was recognised together with a immunodeficiency (multiple food allergies, eosinophilia, eosinophilic gut disease, hyper-IgE, increased GATA3+ T cells). This patient was exome sequenced and no other causative mutation was identified. We therefore investigated a further 20 patients with IL11RA loss of function defects (and skeletal abnormalities indicating defective IL11 signalling) for signs of immunodeficiency. One further patient an immune defect was identified but we did not identify a consistent pattern. We therefore conclude that the IL11 signalling pathway is upregulated during Th17 associated intestinal inflammation (both Crohn's disease and ulcerative colitis) but absent IL11 signalling does not bias the immune response as a high penetrance immunodeficiency Th2 phenotype.

In addition, that time in Oxford allowed me to attend outpatients clinics, the day care/ endoscopy unit and the ward rounds at the Pediatric Gastroenterology Department at the Children's Hospital at the John Radcliffe Hospital. I also joined patient reviews, clinical-histology meetings and teaching sessions. I was lucky enough to understand not only IBD, but also management of genetic disorders, immune deficiency syndromes and very early onset IBD in variety of patients with different problems and complications. This enhanced my clinical experience and contributed to my professional and personal development. The colleagues at the Children's hospital made my visit very efficient sharing their experience and knowledge with me. I enjoyed the friendly atmosphere that gave me the feeling of support. I learned a lot and I am very thankful to Professor Uhlig for supporting me.

I am so grateful to have been awarded this opportunity by ESPGHAN, without which I would not have been able to attend the placement. The grant covered travel expenses and accommodation costs. Thank you indeed!

Yours sincerely, Sema Berktas

