

Current activities ESPN WG Renal Genetics (July 6th 2015)

Board Members:

Elena Levtchenko, Sevgi Mir, Detlef Böckenhauer, Carsten Bergmann, Martin Konrad

Educational Officer:

Djalila Mekahli

Additional members:

19 members from countries throughout Europe (July 2015)

Contact ESPN: Stefanie Weber

Contact EDTA WG: Francesco Emma, Olivier Devuyst

Next meeting of the WG: Sept 3rd 2015, 9:00 a.m. – 11:00 a.m. in Brussels (ESPN meeting)

Past/present activities:

1. "best practise guidelines" for Gitelman syndrome  
Initiative made by Olivier Devuyst, first meeting held in Sept 2013 in Brussels,  
Group members include Elena, Detlef, Francesco, Martin, Dr. Vargas, Dr. Karet  
Current standing: 2nd screening finished, selected publications need now to be mined for  
valuable information that will be used for the guidelines.
2. 1st official meeting of the ESPN Working Group took place in Porto last september, we had  
35 attendants, mainly working group members, the 2nd meeting is planned for this year's  
ESPN in Brussels (Sept 2015)

Several projects from group members were proposed and active participation was encouraged:

- ADPKiD study (F. Emma, F Schaefer), invitation on the website  
>200 patients were enrolled, the study recruitment ended march 2015
- LOWE syndrome, long term follow-up (M. Zaniew, M Ludwig)  
intense discussion about the study design which could be further improved, but general  
agreement to support this effort.
- ARPKD registry (M. Liebau, F. Schaefer)  
Max introduced the registry to the group and invited everybody to collaborate in this  
project. To date, more than 100 patients are registered (Jan 2015)
- Cystinosis (E. Levtchenko)  
Elena introduced the project and invited all group members to collaborate

3. ESPN-WG joined project:  
As discussed at the Porto WG meeting, the WG agreed on a first own project. This project  
aims at the diverse policies concerning living-related kidney transplantation in the context of  
underlying inherited diseases.  
To analyse this in detail, we agreed on a two-step procedure:  
1st step: development of a web-based questionnaire to assess the current strategies  
throughout Europe. Carsten and Martin are currently preparing a 1st draft which next needs  
to be finalized during a first meeting of a sub-committee (including Olivia Boyer, Remi  
Salomon, Carsten Bergmann, Stefanie Weber, Martin Konrad). This committee is still open

for all WG members. This meeting will be held in early summer 2015 and will be financed (in part) by the ESPN (a 2000 Euro grant is provided).

2nd step: data search for topics/items emerging through step 1. This step will be performed in collaboration with the Transplant WG (B.Tönshoff). We think about using data from the EDTA registry and the CERTAIN registry.

#### 4. Survey on genetic testing in ADPKD

Both, the ESPN and the EDTA/ERA WGs have been approached by individual researchers (including Djalila Mekhali, Elena Levtchenko, Franz Schaefer) to support their initiative to define differing attitudes between adult and pediatric nephrologist towards presymptomatic testing of individuals/children at risk for ADPKD.

A questionnaire designed by this research team was sent out to the scientific boards of both groups. At the end, the ESPN WG asked for some changes concerning several specific questions but in general agreed to support this initiative. The EDTA/ERA WG had major concerns mainly related to conflict of interest issues. After a telephone conference in Jan 2015, the WG agreed to proceed as follows:

1. After adaptation of the questionnaire, the questionnaire was sent to the ESPN members to get the opinion of the pediatric community (a 2nd reminder was sent recently).
2. The adult community will be first evaluated in Belgium and Germany as a kind of a pilot study. Afterwards, the analysis of all European countries with the support of the EDTA/ERA WG will be rediscussed.