Report of the ESPN WG on Inherited Renal Disorders (2016)

As part of its activities, the WG has produced in 2016

1. A multicentric retrospective study by Zaniew et al (NDT 2016) comparing 88 patients with Lowe syndrome to 18 patients with Dent 2 disease.
2. The ADPKiDs study has been completed and a manuscript is under preparation (in collaboration with the ERA-EDTA WKIKD)
3. A large cohort of patients with cystinosis has been assembled and a manuscript is expected in the first half of 2017 (sponsored by Orphan Europe)
4. A survey on attitude towards "Living donor kidney transplantation in hereditary nephropathies" has been completed, with nearly 120 responses. Results are being analyzed.
5. Two members of the working group (Licia Peruzzi and Velibor Tasic) will attend in March Paris a 2-day meeting to establish guidelines for Beckwith Wiedemann syndrome. The ESPN WKIRD will sponsor one person and the other will be sponsored by the local organizing committee.

Strategic plans for 2017

On January 29, 2017, the working group committee has met in Leuven (Elena Levtchenko, Detlef Bockenhauer, Carsten Bergmann, Francesco Emma; Martin Konrad (past chair) was excused, Sevgi Mir could not attend) to discuss strategic plans for 2017. Several options were discussed and, based on the limited funds available, the following proposals were made for 2017

1. Four multicentric retrospective studies based on questionnaires:
    a. Nephrogenic Diabetes Insipidus (Detlef Bockenhauer)
    The study will look at long term outcome, including renal function, hydronephrosis, intellectual abilities and growth. It will also analyze treatments. The study will be performed in association of the ERA-EDTA working group on inherited kidney diseases and ERKNet
    b. Bartter and Gitelman (Francesco Emma)
    The study will investigate cross-sectionally the prevalence of hyperparathyroidism in these patients, the renal function at 16-18 years of age (end of pediatric period). Growth charts and therapies will be reviewed.
    c. Comparison of twin patients with cystinosis (Elena Levtchenko)
    The goal is to collect 25 pairs of siblings with cystinosis to see if earlier diagnosis in the second sibling improves the outcome
    d. Collection of cases with proven HNF4A mutations (Martin Konrad)
    The goal is to collect enough cases to describe better the phenotype
2. One expert opinion / guideline paper on X-linked hypophosphatemic rickets, in association with the ERA-EDTA working group on inherited kidney diseases and ERKNet (Dieter Haffner).