

Report of the ESPN Inherited Disorders Working Group

Coordinators: F. Emma; **Board:** D. Bockenhauer, E. Levtchenko, C. Bergman, S. Mir



WORKING GROUP MEETING 2017: January 27, 2017, Leuven

PUBLISHED & SUBMITTED PAPERS:

- The ADPKiDs study completed and is in revision in CJASN (in collaboration with the ERA-EDTA WKIKD)
- Long-term renal outcome in children with OCRL mutations: retrospective analysis of a large international cohort. Zaniew M, Bökenkamp A, Kolbuc M, La Scola C, Baronio F, Niemirska A, Szczepanska M, Bürger J, La Manna A, Miklaszewska M, Rogowska-Kalisz A, Gellermann J, Zampetoglou A, Wasilewska A, Roszak M, Moczko J, Krzemien A, Runowski D, Siten G, Zaluska-Lesniewska I, Fonduli P, Zurrida F, Paglialonga F, Gucev Z, Paripovic D, Rus R, Said-Conti V, Sartz L, Chung WY, Park SJ, Lee JW, Park YH, Ahn YH, Sikora P, Stefanidis CJ, Tasic V, Konrad M, Anglani F, Addis M, Cheong HI, Ludwig M, Bockenhauer D. *Nephrol Dial Transplant.* 2018 Jan 1;33(1):85-94.

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PAPERS IN PREPARATION :

- Cystinosis registry paper is in advanced phase, will be submitted in 2018 (sponsored by Orphan Europe)
- Expert opinion paper on X-linked hypophosphatemic rickets, in association with the ERA-EDTA working group on inherited kidney diseases and ERKNet is in process.
Final meeting took place in December 2017 (Dieter Haffner).

COMPLETED STUDIES:

- Distal RTA questionnaire in association with ERA-EDTA working group on inherited kidney diseases and ERKNet (Detlef Bockenhauer)

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Five multicentric retrospective studies based on questionnaires will be presented in Glasgow:

- **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
- **PTH levels in 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.
- **Sibling 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
- **Cases with proven HNF4A mutations** (Martin Konrad)
The goal is to collect enough cases to describe better the phenotype
- **Cases with contiguous deletions of TSC2 and PKD1** (Carsten Bergmann).
The rationale of this study is based on the limited data available in the literature on, which results in early and severe polycystic kidney disease.
The goal is to collect 50 cases to:
 - describe better the phenotype
 - investigate if all patients are isolated cases within their families
 - to check if all patients are likely due to the same kind of de novo mutation

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DISTAL RTA REGISTRY:

- Contract Advicenne
- dRTA Registry based on AMC platform

PLANNED MEETINGS OF THE WORKING GROUP 2018:

- March 23, 2018 Leuven
- October, 2018 Antalya