Report of the ESPN Inherited Disorders Working Group

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

WORKING GROUP MEETING 2018:
March 23, Leuven

PUBLISHED :

- Prevalence of Hypertension in Children with Early-Stage ADPKD.
  Massella et al. CJASN 2018; 13: 874-833

- Risk Factors for Early Dialysis Dependency in Autosomal Recessive Polycystic Kidney Disease.
  Burgmaier et al. J. Pediatr 2018; 199: 22-28

- Long-term renal outcome in children with OCRL mutations: retrospective analysis of a large international cohort. Zaniew M, Nephrol Dial Transplant. 2018 ;33 :85-94.

- Clinical and molecular diagnosis, screening and management of beckwith-Wiedemann syndrome: an international consensus statement.
  Brioude et al. Nature Rev 2018; 14: 229-249 (Licia Peruzzi represented the Working Group)

- Treatment and long-term outcome in primary distal renal tubular acidosis. Polez-Garcia et al. NDT 2019, February 18

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ACCEPTED PAPERS:

- Clinical practice recommendations for the diagnosis and management of X-linked hypophosphatemia.

  Haffner et al. Accepted for publication in Nature Rev.

COMPLETED STUDIES:

- Cystinosis: siblings study (K Veys et al, paper in preparation)

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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.

- **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 rational of this study is 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 ERKNet platform

PLANNED MEETINGS OF THE WORKING GROUP 2019:

- Not yet planned