european society for paediatric nephrology

WG Inherited Kidney Diseases

Published papers (selection):

- Verploegen MFA et al.. Parathyroid hormone and phosphate homeostasis in patients with Bartter and Gitelman syndrome: an international cross-sectional study. *Nephrol Dial Transplant*.
- Ajiri R et al. Phenotypic Variability in Siblings with Autosomal Recessive Polycystic Kidney Disease. *Kidney International Reports*
- Burballa C et al. Clinical and genetic characteristics of Dent's Disease Type 1 in Europe. *Nephrol Dial Transplant*.
- Groothoff JW et al. Clinical Practice recommendations for primary hyperoxaluria: an expert consensus statement from ERKNet and OxalEurope *Nature Reviews Nephrology*.
- Halawi et al. Clinical Characteristics and Courses of Patients With Autosomal Recessive Polycystic Kidney Disease-Mimicking Phenocopies. *Kidney International Reports*

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Guideline work (selection):

Ongoing/finished:

- Update on XLH, D. Haffner
- NDI (together with ERKNet), N.Knoers/E. Levtchenko
- Dent (together with ERKNet), A. Bokenkamp
- Alport (supporting an ERA-initiated project), R. Torra (ERA), R. Topaloglu for ESPN
- TSC (with ERKNet), D. Mekahli & M. Marlais for ESPN

Planned for 2024:

- update ARPKD (together with ASPN, ERKNet), M. Liebau
- Disorders of Na-Pi transporters, D. Haffner + P. Schlingmann

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Ongoing studies and projects (selection)

- Mutations in the sodium-phosphate cotransporter genes – survey (L. Brunkhorst, D. Haffner)
- The role of Copeptin as a biomarker of volume status in pediatric polyuric tubulopathies. (ESPN research grant). (A. Ferrulli, R. Topaloglu, E. Levtchenko)
- Use of NSAID in patients with tubulopathies (F. Emma, D. Bockenhauer)
- HNF4A survey (P. Schlingmann, F. Emma)
- Pregnancies in ARPKD – with ERKNet & ERA (M. Gosselink, A. van Eerde, M. Liebau)
- Alport syndrome management survey (S. Williamson, R. Lennon)
- Delphi process on "diagnostic and therapeutic management in patients with primary hyperoxaluria" (L. Deesker, J. Groothoff)
- Delphi process on Clinical Practice Recommendations for Dent disease. (A. Bökenkamp)

- dRTA Registry D. Bockenhauer
- ADPKD Registry D. Mekahli
- ARPKD Registry M. Liebau
- Cystinosis Registry A. Servais & E. Levtchenko
- Oxaleurope J. Groothoff

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Educational activities:

- Joint ERKNet-ESPN hybrid course “Innovative Therapies for Rare and Inherited Kidney Diseases” in Heidelberg in November 2022 (~ 400 registrations, ~ 300 participants - from at least 14 countries), supported by ESPN funding

- ESPN WG Webinar “How to diagnose and how to treat hereditary kidney diseases?” in April 2023 (>100 participants)

- ERA-Webinar with ESPN participation in December 2022 (“Genetics and CKD: how common it is? When should we think about it?“ >100 participants, very positive evaluation)

- Planned ESPN-Webinar with ERA participation in autumn 2023 (““Genetic kidney diseases throughout life”)

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Number of members 73

Chair: **Max Liebau**
Board Members: **Gema Ariceta**
George Costea (YPNN)
Dieter Haffner *
Rachel Lennon
Rezan Topaloglu

Liaison to ERA: **Elena Levtchenko**

** Rotating off the board on December 31th, 2023*