Report of ESPN WGIKD Board 2024 (February)

WG composition

Chair: M. Liebau (2022)

Board members: G. Ariceta (2021), R. Lennon (2021), R. Topaloglu (2023), G. Costea (YPNN, 2023), Martine Besouw (2024)

Member of the YPNN: George Costea

Newly elected board member (serving from 1st of January 2024): Martine Besouw

Liaison to ERA: E. Levtchenko

Members: 76 members (+3 in 2023)

Brief description of the scope of the WG

The ESPN working group on inherited kidney diseases aims to improve the care of children with IKD and to foster research on clinical and pathophysiological aspects of IKD in children and adolescents.

Published papers 2023

- Riedhammer et al., "Is there a dominant-negative effect in individuals with heterozygous disease-causing variants in COL4A3/COL4A4?". Clin Genet. 2024 Jan 12. doi: 10.1111/cge.14471.
- Halawi AA et al., „Clinical characteristics and courses of patients with Autosomal Recessive Polycystic Kidney Disease-mimicking phenocopies". Kidney International Reports 2023 Apr 13;8(7):1449-1454.
- Burballa et al., "Clinical and genetic characteristics of Dent's disease type 1 in Europe". Nephrol Dial Transplant. 2023 May 31;38(6):1497-1507. doi: 10.1093/ndt/gfac310.
- Regnier et al., "Worldwide disparities in access to treatment and investigations for nephropatic cystinosis: a 2023 perspective". Pediatr Nephrol. 2023 Nov 18. doi: 10.1007/s00467-023-06179-3. Online ahead of print.

Online ESPN Course 2023

April 2023: "How to diagnose and how to treat hereditary kidney diseases?"

Moderators: Levtchenko, Costea, Cornec LeGall

Talks:

Emma/Bockenhauer: Updates on management of Bartter and Gitelman

Costea: To know or not to know…. Not your typical Dent family

Van Eerde: Look beyond your typical horizon: genetic kidney diseases in adults

Liebau: Concluding remarks

→ More than 100 participants

Webinar together with ERA-EDTA planned for **May 2024: "Genetic kidney diseases throughout life"** (including talk by J. Groothoff and discussion pannelist E. Levtchenko or L. Pape)

Ongoing ESPN-supported registries

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

Ongoing projects

- Glucose-metabolism in early ADPKD: a multi-center study (supported by ESPN research grant). A. Dachy, D. Mekahli
- Mutations in the sodium-phosphate cotransporter genes – survey
  L. Brunkhorst, D. Haffner
- Collecting duct water reabsorption in cystinosis
  A. Ferrulli, G. Valenti, E. Levtchenko
- The role of Copeptin as a biomarker of volume status in pediatric polyuric tubulopathies. study (supported by 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
- Alport Survey
  S. Williamson, R. Lennon
- Pregnancies in ARPKD – with ERKNet, ERA
  M. Gosselink, A. van Eerde, M. Liebau

Requests for support in 2023

- Guideline on Dent's disease, Delphi process/ (together with ERKNet), A. Bokenkamp
- Survey by Detlef Bockenhauer and Francesco Emma on patients with clinical diagnosis of dRTA and the *ATP6V1B1* variant
- Survey by Samantha Williamson and Rachel Lennon on management of Alport's syndrome

Planned/Almost-done New Guidelines & Clinical Practice Recommendations

- XLH update (within this year), D. Haffner
- NDI (together with ERKNet), N.Knoers/E. Levtchenko
- Dent (together with ERKNet), A. Bokenkamp (Delphi process in summer)
- Alport (supporting an ERA-initiated project), R. Torra (ERA), R. Topaloglu for ESPN

- TSC (with ERKNet), D. Mekahli & M. Marlais for ESPN (*in press*, Nat Rev Nephrol)
- update ARPKD (together with ERKNet), M. Liebau (meeting planned for June)
- Disorders of Na-Pi transporters, D. Haffner + P. Schlingmann

Financial report

Aiming to apply for support of the WG meeting for the ARPKD recommendation update

ESPN Board Examination questions on inherited kidney disorders

WGIKD board will participate in writing questions for the next ESPN Board on Inherited Kidney Disorders