Report of ESPN WGIKD Board 2025 (March)

WG composition

Chair: Max Liebau (2022)

Board members: Rezan Topaloglu (2023), George Costea (YPNN, 2023), Martine Besouw (2024), Leire Madariaga Dominguez (2025), Bshara Mansour (2025)

Member of the YPNN: George Costea

Newly elected board members (serving from 1st of January 2025): Leire Madariaga Dominguez, Bshara Mansour

Liaison to ERA: Elena Levtchenko

Members: 78 members (+2 in 2024/2025)

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 2024

- Brunkhorst et al., "Presentation and outcome in carriers of pathogenic variants in SLC34A1 and SLC34A3 encoding sodium-phosphate transporter NPT2a and 2C" Kidney Int. 2025 Jan;107(1):116-129. doi: 10.1016/j.kint.2024.08.035. Epub 2024 Oct 24.
- Gross et al., "SGLT2 inhibitors: approved for adults and cats but not for children with CKD" Nephrol Dial Transplant 2024 Feb 2:gfae029. doi: 10.1093/ndt/gfae029
- 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.
- Regnier et al., "Worldwide disparities in access to treatment and investigations for nephropatic cystinosis: a 2023 perspective". 2024 Apr;39(4):1113-1123. doi: 10.1007/s00467-023-06179-3. Epub 2023 Nov 18.
- Mekahli et al. „Clinical practice recommendations for kidney involvement in tuberous sclerosis complex: a consensus statement by the ERKNet Working Group for Autosomal Dominant Structural Kidney Disorders and the ERA Genes & Kidney Working Group"
- Torra et al., „Diagnosis, management and treatment of the Alport syndrome - 2024 guideline on behalf of ERKNet, ERA and ESPN" Nephrol Dial Transplant. 2024 Dec 2:gfae265. doi: 10.1093/ndt/gfae265.

Online ESPN Course 2024

May 2024: "Recent developments in the field of SGLT2 inhibitors – towards new age groups"

Moderators: Levtchenko, Capasso (ERA)

Talks:

Gross: "Rationale and protocol of a randomized controlled SGLT2 inhibitor trial in pediatric and young adult populations with CKD: DOUBLE PRO-TECT Alport."

Oni: “SGLT2i inhibitors: an example of the challenges in conducting clinical trials in children.”

→ good attendance incl. FDA

November 2024: “Coming of age: inherited kidney diseases throughout life - Primary hyperoxaluria as a case example”

Moderators: Lars Pape, Lucile Figueres (ERA)

Speakers Jaap Groothoff, Sandrine Lemoine (ERA)

Webinar together with ERA-EDTA Februar 2025**: “Kidney Stones”**

ESPN Speaker: Justine Bacchetta

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
- 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, L. Madariaga
- Use of NSAID in patients with tubulopathies, F. Emma, D. Bockenhauer
- HNF4A survey, P. Schlingmann, F. Emma

Requests for support in 2024

- Editorial on electrolyte substitution (together with ERA and ERKNet), ERKNet ePAGs
- Survey by David Csomo on patients with pRTA
- Bone phenotype in aHUS patients on long-term Eculizumab (Justine Bacchetta)
- TheRaCil survey on access to genetic testing (A. Van Eerde)
- Survey on pregnancies in ARPKD (M. Liebau, A. van Eerde)

Planned/Almost-done New Guidelines & Clinical Practice Recommendations

- XLH update (published in 2025, NRN), D. Haffner
- NDI (published in 2025, NRN), N.Knoers/E. Levtchenko
- Dent (published in 2025, NDT), A. Bokenkamp
- update ARPKD (together with ERKNet), M. Liebau (being written)
- Disorders of Na-Pi transporters, D. Haffner + P. Schlingmann
- Transition statement with WG Genes & kidney ERA
- Lowe syndrome (F. Emma)

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