

1st International NephGen Symposium

September 18 - 21, 2023
Historical Merchants' Hall, Freiburg im Breisgau

Monday, September 18, 2023

8.30 - 9.00 am

Registration

9.00 - 9.30 am

Lutz Hein, Anna Köttgen

Welcome & Opening Remarks

9.30 - 10.10 am

Plenary lecture:

Anna Greka

From rare to common: mechanisms of disrupted membrane biology in the kidney and beyond

Session 1

Chairs: Kai Schmidt-Ott, Anna Köttgen

10.10 - 10.40 am

Tobias Hermle

Nephrin endocytosis and cellular dynamics: insights from a *Drosophila* podocyte model

10.40 - 11.10 am

Coffee break

11.10 - 11.40 am

Anna Köttgen

Population-based genetic analyses of complex human kidney traits

11.40 - 11.50 am

Selected short talk: Max Warncke

Immune cell landscaping reveals a protective role for regulatory T cells during kidney injury and fibrosis

11.50 - 12.20 pm

Emilie Cornec-Le Gall

The expanding spectrum of ADPKD

12.20 - 12.50 pm

Felix Knauf

Combining physiology with genetics to better understand tubulorenal diseases

12.50 - 1.50 pm

Lunch break

Session 2

Chairs: Amandine Viau, Carola Hunte

1.50 - 2.20 pm

Jens Leipziger

The molecular physiology of renal base excretion

2.20 - 2.50 pm

Helle Prætorius

Appetite regulating GPR39 agonist reduces renal urinary concentration capacity by interfering with AVP-induced AQP2 trafficking

2.50 - 3.20 pm

Carsten Bergmann

Characterization of new proteins in polycystic kidney disease and related ciliopathies

3.20 - 3.30 pm

***Selected short talk:* Carlotta Pioppini**

Genetic interaction between XBP1 and Pkd1 modulates ADPKD progression

3.30 - 4.00 pm

Coffee break

Session 3

Chairs: Karen Lopez-Cayuqueo, Anke Schumann

4.00 - 4.30 pm

Bernd Fakler

High-resolution complexome of the mouse brush-border membrane

4.30 - 5.00 pm

Dorothee Günzel

Claudinopathies and renal function

5.00 - 5.10 pm

***Selected short talk:* Anke Schumann**

Renal phenotype in a hypomorphic murine model of propionic aciduria

5.10 - 5.40 pm

Carsten Wagner

Phosphate and the kidney

6.30 - 8.00 pm

Historical city tour

Tuesday, September 19, 2023

Session 4

Chairs: Athina Ganner, Christoph Schell

9.00 - 9.30 am

Ian Frew

Recruitment of myeloid cells by tumour cells underlies immune checkpoint therapy resistance in a mouse model of ccRCC

9.30 - 10.00 am

Peter Boor

Deep Learning 4 Nephropathology... and beyond...

10.00 - 10.10 am

***Selected short talk:* Frank Bienaimé**

Primary cilia drive inflammatory responses to urinary flow obstruction and pathogens

10.10 - 10.40 am

Heymut Omran

Genetic defects affecting primary and motile cilia

10.40 - 11.10 am

Coffee break

Session 5

Chairs: Constanze Kainz, Kai-Uwe Eckardt

11.10 - 11.40 am

Elke Neumann-Haefelin

Translational aspects of the VHL research consortium in Freiburg: Establishment and comprehensive characterization of the von Hippel Lindau disease Registry

11.40 - 11.50 am

Selected short talk: Mojca Adlesic

Kif3a heterozygous and homozygous mutations reveal sex-specific differences in the evolution of ccRCC in a mouse model

11.50 - 12.20 pm

Volker Haase

Dissecting oxygen and mitochondrial metabolism in the kidney

12.20 - 12.50 pm

Patient representative presentation:

Louisa Merscher

12.50 - 1.50 pm

Lunch break

Session 6

Chairs: Tobias Huber, Pascal Schlosser

1.50 - 2.20 pm

Christoph Kuppe

Using single cell genomics to dissect mechanisms of kidney diseases

2.20 - 3.00 pm

Plenary lecture:

Katalin Susztak

From kidney function mapping to kidney disease causing genes and cell types

3.00 - 3.30 pm

Coffee break

3.30 - 3.50 pm

Poster Flash I

3.50 - 5.50 pm

Poster Discussion I

7.00 pm

Group dinner

Restaurant "Waldsee"

Wednesday, September 20, 2023

Session 7

Chairs: Julia Höfele, Daniela Braun

9.00 - 9.30 am

Adriana Hung

APOL1 and precision nephrology

9.30 - 10.00 am

Rachel Lennon

Basement membrane genes and kidney phenotypes - a widening spectrum

10.00 - 10.30 am

Catherine Meyer-Schwesinger

Podocyte proteostasis in membranous nephropathy: a creative new concept

10.30 - 10.40 am

Selected short talk: Bayram Edemir

The enriched expression of aquaporin-2 in the renal inner medulla is mediated by increased chromatin accessibility

10.40 - 11.10 am

Coffee break

Session 8

Chairs: Matias Simons, Corinne Antignac

11.10 - 11.40 am

Miriam Schmidts

Implication of FOXD2 dysfunction in syndromic congenital anomalies of the kidney and urinary tract (CAKUT)

11.40 - 12.10 pm

Patient representative presentation II:
Alexander Richter

12.10 - 12.40 pm

Paola Romagnani

Personalized management of steroid-resistant nephrotic syndrome

12.40 - 12.50 pm

Selected short talk: Pascal Schlosser

Integrated proteomic and metabolomic modules associated with risk of kidney function decline

12.50 - 1.50 pm

Lunch break

Session 9

Chairs: Ralph Witzgall, Ulla Schultheiß

1.50 - 2.20 pm

Michael Köttgen

Loss of co-chaperone function causes ADPKD

2.20 - 2.50 pm

Björn Buchholz

Calcium-dependent cyst growth in polycystic kidney disease

2.50 - 3.30 pm

Plenary lecture:
John Maraganore

Advancing RNAi therapeutics for rare and common diseases

3.30 - 4.00 pm

Coffee break

4.00 - 4.20 pm

Poster Flash II

4.20 - 6.20 pm

Poster Discussion II

7.00 pm

Speakers' dinner
Restaurant "Der Kaiser"

Thursday, September 21, 2023

9.00 - 10.00 am

Round table talk with the editor:

Olivier Devuyst, Editorial Board, Kidney International

Session 10

Chairs: Ian Frew, Stefan Haug

10.00 - 10.10 am

Selected short talk: Stefan Haug & Oleg Borisov

Characterization of the urine proteome in 769 patients with early-stage chronic kidney disease

10.10 - 10.40 am

Thomas Benzing

Molecular design of the kidney filtration barrier

10.40 - 11.10 am

Coffee break

11.10 - 11.40 am

Matthias Wuttke

Large-scale exome sequencing studies highlight kidney-function related genes and variants

11.40 - 11.50 am

Selected short talk: Ioanna Tachmazidou

Whole-exome sequencing analysis identifies genes associated with kidney function and damage in multi-ancestry participants of the UK Biobank

11.50 - 12.30 pm

Plenary lecture:
Melissa Little

Applying kidney organoids to disease modelling and drug development

12.30 - 12.45 pm

Anna Köttgen

Closing remarks

12.45 pm

Lunch and farewell

Poster Discussion I - 19.09.2023

ID #	Author	Title
100	Karl Kunzelmann	Modulation of inflammation by TMEM16J/ANO9
101	Pascal Schlosser	Integrated Proteomic and Metabolomic Modules Associated with Risk of Kidney Function Decline
102	Stefan Haug	Characterization of the Urine Proteome in 769 Patients with Early-Stage Chronic Kidney Disease
103	Oleg Borisov	Colocalization of genetic associations with plasma and urine proteins provide insights into renal protein handling and related clinical outcomes.
104	Nora Scherer	Exome sequencing reveals genes shaping the plasma and urine metabolome and connections to human diseases
105	Hassan Saei	Genotyping coding-VNTR in the MUC1 gene using k-mer frequency based alignment-free method allows genetic diagnosis of MUC1-related autosomal dominant tubulointerstitial kidney disease
106	Jessica Kachmar	Steroid-Resistant Nephrotic Syndrome due to NPHS2 Variants is Not Associated with Post-Transplant Recurrence
107	Matias Simons	Targeting RNF145 in renal proximal tubular cells protects against lipotoxicity in diabetes
108	Anna Klawonn	Deciphering the role of the ciliary Intraflagellar Transport (IFT)- dynein-complex mediated cell signaling network for vertebrate development
109	Julian Milosavljevic	Modeling TBC1D8B-associated FSGS in Drosophila nephrocytes.
110	Dominik Spitz	mTor-dependent autophagy regulates slit diaphragm density in podocyte-like Drosophila nephrocytes
111	Björn Neubauer	Loss of co-chaperone function causes ADPKD
112	Miriam Schmidts	IFT74 variants cause reno-skeletal ciliopathy and motile cilia defects in mice and humans
113	Niklas Ayasse	Trimethoprim inhibits renal H ⁺ /K ⁺ ATPase in states of K ⁺ deprivation
114	Toma Antonov Yakulov	Investigating genetic compensation and robustness in nephronophthisis using zebrafish models
117	Max Warncke	Immune cell landscaping reveals a protective role for regulatory T cells during kidney injury and fibrosis
118	Haihua Guo	ADP-ribosylation factor interacting protein 2 acts as a novel regulator of mitophagy and autophagy in podocytes in diabetic nephropathy
119	Magali Ferro	Kidney organoid to model podocytopathies and identify novel therapeutic targets
120	Konrad Lang	The distinctive functions of endocytic mechanisms within the maintenance of slit diaphragms in Drosophila nephrocytes
121	Céline Schaeffer	Dissecting heterogeneity and common pathogenetic pathways in Autosomal Dominant Tubulointerstitial Kidney Disease due to mutations in REN

Poster Discussion I - 19.09.2023

ID #	Author	Title
122	Antonella Catalano	Vhl and Rassf1a tumour suppressors cooperatively maintain genomic stability in ccRCC
123	Max Bergen	Calcium signaling drives actin network formation in multiciliated cells
124	Marc Brettel	Stable genetic deletion of Gapvd1 in Drosophila results in a nephrocyte-restricted phenotype
125	Lukas Westermann	Analysis of Pkd1 gene function in a 3D tubuloid assay
126	Marc Timmers	Epigenetic control in VHL-dependent clear cell renal cell carcinoma
127	Shuang Cao	Single cell CRISPR screening as a powerful tool for gene regulatory networks studies in kidney collecting ducts
128	Andre Kraus	Knockout of P2Y2R inhibits cyst growth in an ADPKD mouse model
129	Ioanna Tachmazidou	Whole-exome sequencing analysis identifies genes associated with kidney function and damage in multi-ancestry participants of the UK Biobank
130	Tobias Staudner	Generation und electrophysiological analysis of a gain-of-function polycystin-1/polycystin-2 heteromeric ion channel
131	Pascal Kleindienst	Novel interacting proteins of ciliary TZ localized CBY1, TMEM218 and PATJ in PKD and ciliopathies
132	Sahar Ghasemi	Unraveling the Genetic Basis of CKD: Insights from CKDGen Round 5 GWAS Project
134	Henrike Berns	WNT11 loss of function results in renal hypodysplasia, complex heart defects, and laterality defects in humans
135	Carlotta Pioppini	Genetic interaction between XBP1 and Pkd1 modulates ADPKD progression
140	Evgeny V. Mymrikov	Structural basis for the specific interaction between scaffold protein PDZK1 and ion transporter URAT1
149	Florian Becker	Calcineurin B homologous protein 3 anchors the regulatory domain of human sodium/proton exchanger 1 to the membrane through novel target-myristoyl switch

Poster Discussion II - 20.09.2023

ID #	Author	Title
133	Frederic Arnold	Unravelling autoimmunity and tissue residency in a model of CD8 T cell-driven nephritis
136	Miriam Schmidts	Investigating the role of epigenetic factors in congenital anomalies of the kidney (CAKUT)
137	Jagriti Pal	Nonstop extension mutations inactivate tumor suppressor genes in ccRCC
138	Luise Hennicke	Mouse Models of Familial Hyperaldosteronism
141	Constantin Dickel	Stem-cell derived proximal tubule cells as a novel model to study chloride-oxalate transport
142	Peter Walentek	Tra2b – a new paradigm in coordinated post-transcriptional control of ciliogenesis
143	Clara Naomi Thomas	Short-chain-fatty acid Butyrate inhibits intestinal oxalate transport in Caco2 cells
144	Sarah Wendlinger	Establishing methods for renal base editing in-vivo
145	Caroline Maria Ebersbach	The role of Wnt signaling in distal nephron development and autosomal-recessive polycystic kidney disease
146	Shruti Naik	The Gb3 paradox in proximal tubular injury
147	Claire Leroy	A nephrocyte model for the glomerular basement membrane
148	Lea Gerstner	Genome sequencing improves diagnostic yield in genetic kidney disease
150	Mojca Adlesic	Kif3a heterozygous and homozygous mutations reveal sex-specific differences in the evolution of ccRCC in a mouse model
151	Salómon Christer	A Drosophila model for Dent's disease unveils a role for chloride/proton exchanger Clc-c in regulating the cortical actin cytoskeleton
152	Janek Jonathan Haus	Novel Candidate Disease Gene for Familial Hyperaldosteronism
153	Petra Stöbe	Tackling undiagnosed patients by long-read genome sequencing
154	Kathrin Skoczynski	Potential relevance of ATP and calcium-mediated chloride secretion via P2Y2R and TMEM16A in human ADPKD kidneys
155	Annika Merz	Role of cell-ECM interactions for renal tubular epithelial cell phenotypes in the context of progressive chronic kidney disease
156	Miriam Schmidts	Prostaglandin synthesis defects resulting from PLA2G4A loss of function causes a phenotype resembling NSAID-nephropathy
157	Maximilian Wess	Subcellular localisation patterns of ANXA4 influence migratory modes in ccRCC
158	Athina Ganner	Elucidating new mechanisms of mTOR regulation in VHL-dependent clear cell renal cell carcinoma
159	Manuel Rogg	Methodological toolbox for the analysis of the cancer matrisome in urogenital malignancies

Poster Discussion II - 20.09.2023

ID #	Author	Title
160	Gerlineke Hawkins-van der Cingel	A triple threat to tubular phosphate reabsorption
161	Amandine Viau	Primary cilia drive inflammatory responses to urinary flow obstruction and pathogens
162	Jefferson Lorenzo Triozzi	Genetic proxies of thiazide diuretics lower the risk of kidney stones in a Mendelian randomization meta-analysis
163	Jefferson Lorenzo Triozzi	Accelerating Drug Discovery for CKD and ESKD through Mendelian Randomization Analyses of Druggable Proteins
164	Tobias Feilen	Proteomic signatures in sporadic progressive and non-progressive clear cell renal cell carcinoma
165	Frank Bienaimé	Basement membrane remodelling links cilia signaling to tubule mechanics in autosomal dominant polycystic kidney disease
166	Constanze Kainz	Contribution of obesity to the initiation and promotion of clear cell renal cell carcinoma
167	Manuel Andreas Anderegg	Prevalence and characteristics of monogenic disease in adult kidney stone formers
168	Todd Alexander	Clinical and Molecular Characterization of Monoallelic and Biallelic SLC34A3 c.1248_1249del Variants
169	Jasmina Ćomić	Exome sequencing in a large cohort of individuals with VATER/VACTERL association as a modern tool to identify phenocopies
170	Edemir Bayram	The enriched expression of aquaporin-2 in the renal inner medulla is mediated by increased chromatin accessibility
171	Dariusz Ghasemi-Semeskandeh	Phenome-wide, metabolomic and proteomic association scan of SHROOM3 haplotypes based on imputed exonic variants