

FLORENCE 2025 20-22 MARCH



Metabolic bone disease and cardiovascular health in kidney disorders

PROGRAMME



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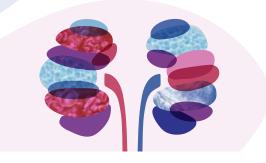
Scientific Programme

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ESPN RESEARCH CONFERENCE FLORENCE 2025 20-22 MARCH



Dear colleagues and dear friends,

We are excited to host you in the 1st ESPN Research Conference from March 20-22, 2025. During the next 2 days, scientists and clinicians are going to share unrivaled research and inspiring new ideas on the interplay of bones, kidneys, and cardiovascular system in health and disease.

The program ensures extensive discussions during the scientific sessions and networking activities to advance research ideas, promote collaboration, and establish the culture innovation.

We have reserved a large space for free oral presentations and posters allowing young and more experienced researchers to present and discuss their latest work with experts in the field.

Let's enjoy discussion, exchange ideas and gain insights into disease mechanisms and effects of novel treatments in a stimulating retreat atmosphere!

Thank you for coming to Florence!

Prof. Dieter Haffner ESPN President PD Dr. Maren Leifheit-Nestler Scientific Co-Chair

Prof. Stella Stabouli Scientific Co-Chair

M. Keifheit-Nestlo SS pung -



THURSDAY, 20 MARCH 2025

14:00 Arrival at the Hotel Mediterraneo and check-in

- **16:00** Welcome from the ESPN President **Dieter Haffner** (Hannover Medical School, Germany)
- 16:05 Introduction from the Chairs
 Stella Stabouli (Aristotle University of Thessaloniki, Greece) and
 Maren Leifheit-Nestler (Hannover Medical School, Germany)
- **16:10** KEYNOTE SESSION
- 17:30 Discussion Leaders: Stella Stabouli (Aristotle University of Thessaloniki, Greece) and Maren Leifheit-Nestler (Hannover Medical School, Germany)
- 16:10 Phosphate homeostasis and endocrine regulators
 Clemens Bergwitz (Yale University School of Medicine, USA)
- 16:40 Discussion
- 16:50 Understanding of metabolic bone diseases: lessons from genetic animal models Carsten Wagner (University of Zurich, Switzerland)
- 17:20 Discussion
- 17:30 POSTER SESSION 1
- **18:30** Coffee will be served during the poster session

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THURSDAY, 20 MARCH 2025

- 18:30 DIAGNOSIS AND MANAGEMENT OF CHILDREN AND ADULTS WITH X-LINKED HYPOPHOSPHATEMIA: WHAT'S NEW? + BEST ABSTRACTS
- **20:00** Discussion Leaders: **Dieter Haffner** (Hannover Medical School, Germany) and **Jun Oh** (University Medical Center Hamburg-Eppendorf, Germany)
- 18:30 New XLH European Guidelines: evolving clinical practice
 Dieter Haffner (Hannover Medical School, Germany) and
 Francesco Emma (Bambino Gesù Children's Hospital, Rome, Italy)
- 19:15 Discussion
- 19:30 Markers of metabolic bone disease in children with X-linked hypophosphatemia: a real-world study in Germany and Switzerland Ineke Böckmann (Hannover Medical School, Germany)
- 19:40 Discussion
- 19:45 Naturally occurring stable calcium isotopes predict changes in bone (de)mineralisation in children and young adults with CKD and on dialysis Rukshana Shroff (UCL Great Ormond Street Hospital and Institute of Child Health, London, United Kingdom)
- 19:55 Discussion
- 20:00 Dinner at the hotel's restaurant



FRIDAY, 21 MARCH 2025

07:30 *Breakfast* **08:45**

MEETING ROOM SALONE DEGLI OCEANI

	MEETING ROOM SALONE BEGEF OCEA
_	CARDIOVASCULAR SYSTEM IN KIDNEY DISEASES Discussion Leaders: Dieter Haffner (Hannover Medical School, Germany) and Marc Vervloet (Radboud University, The Netherlands)
09:00	Cardiovascular biomarkers: FGF23, klotho and beyond (cause or marker) Justine Bacchetta (<i>University of Lyon, France</i>)
09:20	Discussion
09:30	Functional analyses of the calcium-sensing receptor (CaSR) in the cardiac system and the impact of high phosphate Louise Baer (Hannover Medical School, Germany)
09:40	Discussion
09:45	Cardiovascular health in CKD: addressing the gaps Rukshana Shroff (Great Ormond Street Institute of Child Health, University College London, UK)
10:05	Discussion
10:15	High phosphate diet-induced cardiac dysfunction: importance of sodium-dependent phosphate cotransporters Pit1 and Pit2 Andrea Grund (Hannover Medical School, Germany)
10:25	Discussion

10:30 Coffee break

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FRIDAY, 21 MARCH 2025

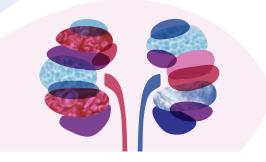
PATHOPHYSIOLOGICAL INSIGHTS INTO METABOLIC BONE DISEASES Discussion Leaders: Justine Bacchetta (University of Lyon, France) and Clemens Bergwitz (Yale University School of Medicine, USA)
Insight of the development of myopathy and future treatments Philippos Mourikis (Necker Institute for Sick Children, France)
Discussion
Bone and vessel crosstalk Marc Vervloet (Radboud University, The Netherlands)
Discussion
Cystinosis metabolic bone disease: pathogenesis and outcome Dieter Haffner (Hannover Medical School, Germany)
Discussion
Group photo
Lunch at the hotel's restaurant
Free time



FRIDAY, 21 MARCH 2025

	NOVEL AND FUTURE THERAPEUTIC MEASURES FOR METABOLIC BONE DISEASE AND CARDIOVASCULAR SYSTEM Discussion Leaders: Maren Leifheit-Nestler (Hannover Medical School, Germany) and Franz Schaefer (University of Heidelberg, Germany)
15:00	Novel measures for metabolic bone disease: PH1-3 and beyond Michiel J.S. Oosterveld (University of Amsterdam, The Netherlands)
15:20	Discussion
15:30	Impact of the enzyme replacement therapy, INZ-701, in children with ENPP1 deficiency: experience from an expanded access program Mattia Parolin (University of Padua, Italy)
15:40	Discussion
15:45	The heart in Fabry disease Kristina Sonnenschein (Hannover Medical School, Germany)
16:05	Discussion
16:15	Musclin as a new marker for muscle health in nephropathic cystinosis Malgorzata Szaroszyk (Hannover Medical School, Germany)
16:25	Discussion
	POSTER SESSION 2 Coffee will be served during the poster session

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FRIDAY, 21 MARCH 2025

MEETING ROOM SALONE DEGLI OCEANI

	EXPERIMENTAL TRANSLATIONAL STUDIES I Discussion Leaders: Michiel J.S. Oosterveld (University of Amsterdam, The Netherlands) and Rukshana Shroff (Great Ormond Street Institute of Child Health, University College London, UK)
18:00	Understanding the pathophysiological signaling in cardiovascular disease Jakob Völkl (Johannes Kepler University Linz, Austria)
18:20	Discussion
18:30	Development and validation of a vascular organoid model for pediatric chronic kidney disease-associated vascular remodeling and calcifications Julie Bernardor (University Hospital of Nice, France)
18:40	Discussion
18:45	Mitochondrial involvement in cardiac, muscle and kidney disease Francesco Emma (Bambino Gesù Children's Hospital, Italy)
19:05	Discussion
19:15	Peritoneal infusion of microplastics induces over-expression of GDF15 in kidney and heart in mice Marta Ferrecchi (University of Genoa, Italy)
19:25	Discussion
<u>19:30</u> 20:00	Free time

20:30 Dinner at "Finisterrae" restaurant



SATURDAY, 22 MARCH 2025

07:30	Breakfast
08:45	

	CLINICAL SCENARIOS Discussion Leaders: Oh Jun (University Medical Center Hamburg-Eppendorf, Germany) and Stella Stabouli (Aristotle University of Thessaloniki, Greece)
09:00	Clinical scenarios in pediatric kidney diseases with a focus on cardiovascular and bone health Franz Schaefer (University of Heidelberg, Germany)
09:20	Discussion
09:30	Longitudinal relationship between albuminuria and renal function in children and young adults from the general population Valentina Gracchi (Beatrix Children's Hospital - University Medical Center Groningen, The Netherlands)
09:40	Discussion
09:45	Clinical scenarios in adults with CKD and cardiovascular outcomes Beatriz Fernández-Fernández (IIS-Fundacion Jimenez Diaz UAM, Madrid, Spain)
10:05	Discussion
10:15	Vitamin K2 profile in young patients with renal replacement therapy: link with vascular health? Lou Cryns (University of Antwerp, Belgium)
10:25	Discussion
10:30 11:00	Coffee break

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SATURDAY, 22 MARCH 2025

	EXPERIMENTAL TRANSLATIONAL STUDIES II Discussion Leaders: Jakob Völkl (Johannes Kepler University Linz, Austria) and Carsten Wagner (University of Zurich, Switzerland)
11:00	Genetically modified animal models Guillaume Courbon (INSERM, University of Saint Etienne, France)
11:20	Discussion
11:30	Identification of phenotype modifier gene variants overrepresented in familial hypomagnesemia with hypercalciuria and nephrocalcinosis with a more aggressive renal phenotype Julieta Torchia (Vall d'Hebron Institute of Research, Barcelona, Spain)
11:40	Discussion
11:45	Genome editing (CRISPR/Cas and others) in kidney diseases Miguel A. Garcia González (University of Santiago de Compostela, Spain)
12:05	Discussion
12:15	Applying LNP/mRNA technology to therapy of cystinosis Paul Goodyer (Research Institute of the McGill University Health Centre, Montreal, Canada)
12:25	Discussion
12:30 12:45	Farewell
<u>12:45</u> 14:00	Luch at the hotel's restaurant



- Po1 Naturally occurring stable calcium isotopes predict changes in bone (de) mineralisation in children and young adults with CKD and on dialysis

 Alexander D. Lalayiannis¹, Anton Eisenhauer², Ana Kolevica², Alexander Heuser², Varvara Askiti³, Amrit Kaur¹, Manish Sinha¹, David Milford¹, Mary Fewtrell¹, Rukshana Shroff¹ (¹United Kingdom, ²Germany, ³Greece)
- Po2 Fanconi-Bickel syndrome: a rare cause of pRTA with significant risk of bone fractures and deformities

 Bahriye Atmis, Aysun Karabay Bayazıt, Deniz Kor, Fatma Derya Bulut,
 Neslihan Önenli Mungan (Turkey)
- Po3 Secondary hyperparathyroidism and brown tumors in pediatric dialysis patients: a critical call for treatment adherence

 Bahriye Atmis, Zahide Orhan Ok, Çağla Çağlı Pişkin, Aysun Karabay Bayazıt

 (Turkey)
- Po6 Alterations of bone and mineral metabolism during CKD progression and impact of kidney replacement therapy: a real-world pediatric study

 Anna Tschirner, Weber Hannah, Schermuly Katharina, Böckmann Ineke,
 Kanzelmeyer Nele, Drube Jens, Haffner Dieter, Leifheit-Nestler Maren (Germany)
- Po7 Markers of metabolic bone disease in children with X-linked hypophosphatemia: a real-world study in Germany and Switzerland

 Ineke Böckmann, Maren Leifheit-Nestler, Schlingmann Karl Peter,
 Freiberg Clemens, Weitz Marcus, Dunstheimer Desiree, John Ulrike, Wühl Elke,
 Schnabel Dirk, Haffner Dieter (Germany)
- Pog Functional analyses of the calcium-sensing receptor (CaSR) in the cardiac system and the impact of high phosphate

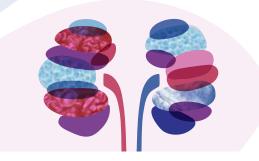
 <u>Louise Baer</u>, Andrea Grund, Dieter Haffner, Maren Leifheit-Nestler (Germany)
- P10 High phosphate diet-induced cardiac dysfunction: Importance of sodium-dependent phosphate cotransporters Pit1 and Pit2

 Andrea Grund, Maren Leifheit-Nestler, Dieter Haffner (Germany)
- P-cresol and p-cresol sulphate and clinical outcomes in CKD: systematic review Patrick Clarke, Matthew J. Harmer (United Kingdom)
- P13 Impact of the enzyme replacement therapy, INZ-701, in children with ENPP1 deficiency: experience from an expanded access program

 Mattia Parolin¹, Belen Ferrer², Ruba Rizik³, Kurt C. Gunter⁴ (¹Italy, ²Spain, ³Israel, ⁴USA)
- P14 Can oral bisphosphonates counteract mineral bone disease in Lowe syndrome? A case series of three patients

 Elisa Manieri, Guido Zavatta, Federico Baronio, Andrea Pasini, Claudio La Scola (Italy)

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- P15 Musclin as a new marker for muscle health in nephropathic cystinosis

 Malgorzata Szaroszyk, Lars Leemhuis, Dieter Haffner, Maren Leifheit-Nestler

 (Germany)
- P16 Developing an organoid model of podocin-related nephrotic syndrome using patient-derived induced pluripotent stem cells

 Robert Matthes, Mansoureh Tabatabaeifar, Sabine Jung-Klawitter, Franz Schaefer (Germany)
- P18 Applying LNP/mRNA technology to therapy of cystinosis Paul Goodyer, Elena Torban (Canada)
- P19 Development and validation of a vascular organoid model for pediatric chronic kidney disease-associated vascular remodeling and calcifications

 Matthieu Rouleau¹, Justine Simonin¹, Elise Larché¹, Zakariya Caillot¹,

 Claudine Blin-Wakkach¹, Maria Bartosova², Claus Peter Schmitt², Julie Bernardor¹

 (¹France, ²Germany)
- P20 Effects of exercise in chronic kidney disease: crosstalk between muscle and kidney Katharina Maier, Malgorzata Szaroszyk, Dieter Haffner, Maren Leifheit-Nestler (Germany)
- Peritoneal infusion of microplastics induces over-expression of GDF15 in kidney and heart in mice

 Marta Ferrecchi, Decimo Silvio Chiarenza, Micaela Gentile, Paolo Cravedi,
 Gabriele Mortari, Pasquale Esposito, Daniela Verzola, Enrico Verrina,
 Noemi Rumeo, Edoardo La Porta (Italy)
- P22 Identification of phenotype modifier gene variants overrepresented in familial hypomagnesemia with hypercalciuria and nephrocalcinosis with a more aggressive renal phenotype

 <u>Julieta Torchia</u>, Mónica Vall-Palomar, Jordi Morata, Monica Duran, Mireia Ferrer, Alex Sanchez, Gerard Cantero-Recasens, Gema Ariceta, Anna Meseguer, Cristina Martínez (Spain)
- P23 Coronavirus viroporins regulate CFTR trafficking and function

 <u>Aparna Renigunta</u>, Jonathan Schlegel, John Ziebuhr, Vijay Renigunta,

 Stefanie Weber (Germany)
- p-glycoprotein-1(abcb1) along with adenylate kinase (ak1) mediates nucleotide -gated channel regulation of kcnj1 Vijay Renigunta¹, Imran Shaikh¹, Nils Renfordt¹, Siegfried Waldegger², Martin Koemhoff¹, Stefanie Weber¹, Aparna Renigunta¹(¹Germany, ²Austria)
- P25 Biomarkers predictive for the renal graft survival Romanian experience Andreea Liana Bot (Rachisan) (Romania)



- P28 Vitamin K2 profile in young patients with renal replacement therapy: link with vascular health?

 Lou Cryns¹, Van Eyck Annelies¹, Leon Schurgers², Dominique Trouet¹ (¹Belgium, ²The Netherlands)
- P29 Variant analysis in a cohort of pediatric patients with congenital anomalies of the kidney and urinary tract (CAKUT) after extended next generation sequencing (NGS)

 Dana Blasi, Mansoureh Tabatabaeifar, Christian Sutter, Franz Schaefer (Germany)
- P30 Insulin resistance in young patients with autosomal dominant polycystic kidney disease Olil Van Reeth, Inès Vanderheyden, Jean-Paul Decuypere, Rudi Vennekens, Peter Janssens, François Jouret, Djalila Mekahli (Belgium)
- P31 Longitudinal relationship between albuminuria and renal function in children and young adults from the general population

 Valentina Gracchi¹, Hiddo Lambers Heerspink², Henk Groen³, Richard Coward², Henkjan Verkade¹ (¹The Netherlands, ²United Kingdom)
- P32 Pediatric nephrolithiasis-clinical manifestations and metabolic profiles from a dedicated multidisciplinary clinic

 Shelly Levi, Roy Morag, Daniel Landau, Miriam Davidovits, Abigail Lazar, Lotem Goldberg, Efrat Talgam Horshi, Gili Koren, Adi Chezana, Yael Borovitz (Israel)
- P33 Percutaneous kidney biopsy associated risks in children: a 10 years clinical experience <u>Donatella Simone</u>, Diletta Domenica Torres, Vincenza Carbone, Luisa Santangelo, Marida Martino, Mario Giordano, Paolo Giordano (Italy)
- P34 Evaluation of carnitine concentration in blood and urine in children with diseases of the kidneys and urinary tract

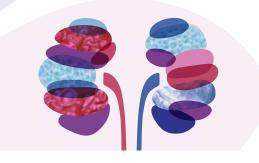
 <u>Agnieszka Jędzura</u>, Paulina Wysocka-Wojakiewicz, Martyna Jasielska,

 Anna Rokowska-Oleksa, Monika Dębowska, Katarzyna Sedlaczek,

 Piotr Adamczyk (*Poland*)
- P35 Pediatric reference values for novel urinary biomarkers of kidney health in the HARP cohort

 Hannah Weber, Katharina Schermuly, Anna Tschirner, Ineke Böckmann,
 Ulrich Baumann, Anibh M. Das, Nele Kanzelmeyer, Jens Drube, Dieter Haffner,
 Maren Leifheit-Nestler (Germany)
- P36 Office blood pressure and obesity in children with X-linked hypophosphatemia Ineke Boeckmann, Maren Leifheit-Nestler, Mirko Rehberg, Marcus Weitz, Ulrike John, Ludwig Patzer, Miroslav Zivicnjak, Elke Wühl, Dirk Schnabel, Dieter Haffner (Germany)

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- P37 Long-term follow-up of patients with cyp24a1, slc34a1 and slc34a3 mutations: a retrospective study

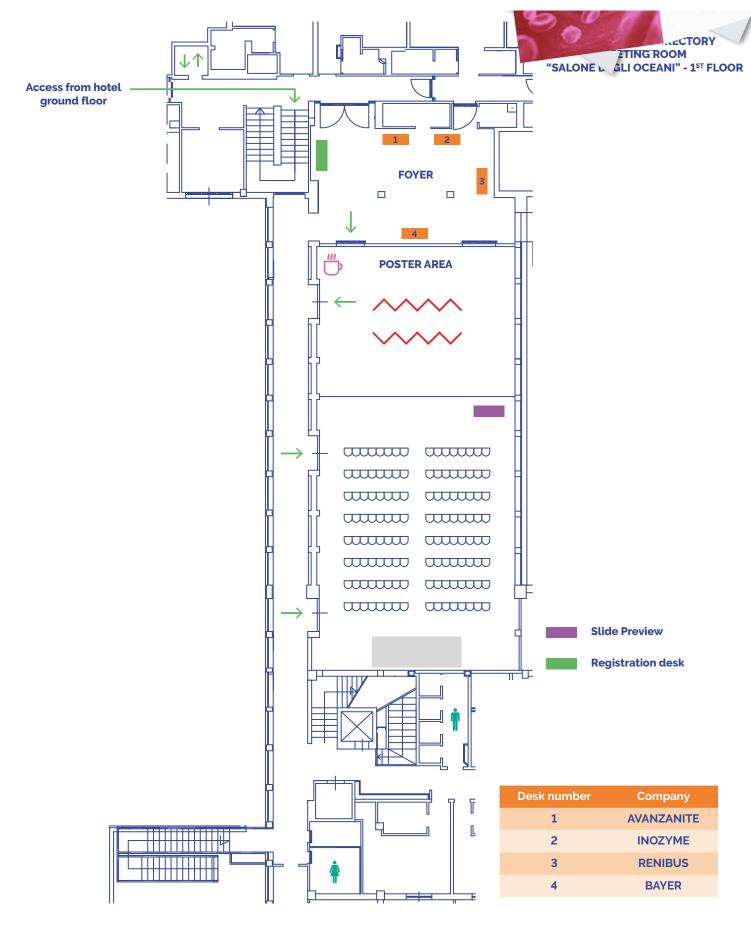
 Ruth Schreiber, Asaf Lebel, Shelly Levi (Israel)
- P38 Obesity as a risk factor for kidney injury in a cohort of children in treatment for acute lymphoblastic leukemia: a multicenter retrospective analysis

 Miriana Guarino, Paolo Giordano, Mario Giordano, Letizia Pomponia Brescia,
 Nicola Santoro, Francesco Chiarelli, Valerio Cecinati (Italy)
- P39 Impact of mineral bone disease on quality of life in children with stage 5 chronic kidney disease

 Branko Lutovac¹, Brankica Spasojevic² (¹Montenegro, ²Serbia)
- P40 Regulation of serum parameters of renal osteodystrophia and hypertension in pediatric patients on chronic hemodialysis

 <u>Kristina Matijas</u>, Mirjana Cvetkovic, Dusan Paripovic, Gordana Milosevski-Lomic, Ivana Gojkovic, Ana Petrovic, Milica Vukanovic, Tanja Gaus, Brankica Spasojevic (Serbia)









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NOTES



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Distal renal tubular acidosis is a rare kidney disorder that occurs when the kidneys are unable to effectively remove the buildup of circulating acids in the blood—a condition called acidosis—which leads to a metabolic imbalance. dRTA is a serious disorder, especially for paediatric patients. dRTA can be either genetically inherited or acquired. The inherited form of dRTA, also known as primary dRTA, is most common among infants and children, while the acquired form is more common in adults. Acquired dRTA can be due to autoimmune disorders, rheumatoid arthritis, or to certain medications. An estimated 30,000 patients in Europe and 20,000 patients in the United States suffer from dRTA.





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RNAi, ribonucleic acid interference.

References:

- 1. Alnylam Pharmaceuticals. Our science. Accessed February 2025. https://www.alnylam.com/our-science/the-science-of-rnai/
- 2. Alnylam Pharmaceuticals. Our pipeline. Accessed February 2025. https://www.alnylam.com/alnylam-rnai-pipeline

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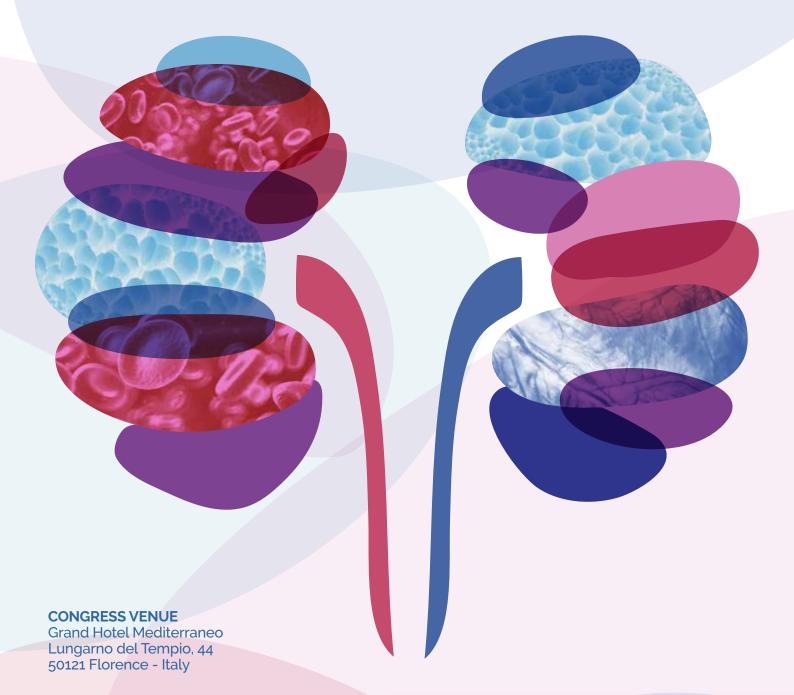












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