International Conference:

The kidney in genetic and rare diseases

Organized by

Prof. Giovambattista Capasso

II University of Naples

biogem

Promoted by

FONDAZIONE INTERNAZIONALE MENARINI

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Symposia: 297

Naples October 27-29, 2016

International Conference: The kidney in genetic and rare diseases

Patients affected by so called *rare diseases* should be able to receive appropriate diagnosis, high quality service, treatment, and support.

Most rare diseases are genetic, and thus are present throughout the person's entire life, even if symptoms do not appear immediately. Therefore, an early diagnosis is crucial for the prompt and effective intervention. Nowadays, thanks to the enormous progress in the field of medical genetics, identification of disease-causing genes and their abnormalities became a routine and effective diagnostic procedure. This is also true for the rare genetic diseases with a manifestation of a prominent renal phenotype.

The main aim of the conference is to shad light on renal genetic and rare disorders as well as to explore "bench-to-bedside" approach, which translates recent genetic and molecular discoveries into clinical settings. In order to create a multidisciplinary platform we assembled an outstanding and highly qualified faculty composed of expert geneticist, molecular biologists, chemists, physiologists, and clinicians.

Taking as a whole, the conference will highlight that a deep understanding of mechanisms behind rare renal diseases leads to better identification of basic mechanism governing renal physiology and pathophysiology.

Prof. Giovambattista Capasso
President of the Conference

President of the Conference

Giovambattista Capasso - *Naples, Italy*

Faculty

Generoso Andria - *Naples, Italy*
Mustafa Arici - *Ankara, Turkey*
Andrea Ballabio - *Naples, Italy*
Olivia Boyer - *Paris, France*
Michael Joseph Caplan - *New Haven (CT), USA*
Santina Castellino - *Catania, Italy*
Mei Changlin - *Shanghai, China*
Annamaria Colao - *Naples, Italy*
Giuseppe Conte - *Naples, Italy*
Salvatore Coppola - *Caserta, Italy*
Antonio Dal Canton - *Pavia, Italy*
Mario De Felice - *Naples, Italy*
Olivier Devuyst - *Zurich, Switzerland*
Vincenzo Di Marzo - *Naples, Italy*
Dominique Eladari - *Paris, France*
Francesco Emma - *Rome, Italy*
Ana Carina Ferreira - *Lisbon, Portugal*
Brunella Franco - *Naples, Italy*
Gerardo Gamba - *Mexico City, Mexico*
Giovanni Gambaro - *Rome, Italy*
Giacomo Garibotto - *Genoa, Italy*
John Geibel - *New Haven (CT), USA*
Loreto Gesualdo - *Bari, Italy*
Maddalena Gigante - *Bari, Italy*
Giuseppe Grandaliano - *Foggia, Italy*
Philippe Jaeger - *Geneva, Switzerland*
Sabina Jelen - *Ariano Irpino, Italy*
Vivekanand Jha - *New Dheli, India*
Katsumasa Kawahara - *Tokyo, Japan*
Moshe Levi - *Denver (CO), USA*
Francesca Mallamaci - *Reggio Calabria, Italy*
Giancarlo Marinangeli - *Giulianova, Italy*
Gennaro Marino - *Naples, Italy*
Paolo Menè - *Rome, Italy*
Piergiorgio Messa - *Milan, Italy*
Orson Moe - *Dallas (TX), USA*
Giovanni Montini - *Milan, Italy*
Luisa Murer - *Padua, Italy*
Vincenzo Nigro - *Naples, Italy*
Marina Noris - *Bergamo, Italy*
Antonello Pani - *Cagliari, Italy*
Norberto Perico - *Bergamo, Italy*
Alessandra Perna - *Naples, Italy*
Francesco Pesce - *Bari, Italy*
Claudio Pisano - *Ariano Irpino, Italy*
Qi Qian - *Rochester (MN), USA*
Teresa Rampino - *Pavia, Italy*
Giuseppe Remuzzi - *Bergamo, Italy*
Dario Roccatello - *Turin, Italy*
Paola Romagnani - *Florence, Italy*
Claudio Ronco - *Vicenza, Italy*
Pierre Ronco - *Paris, France*
Margherita Ruoppolo - *Naples, Italy*
Francesco Salvatore - *Naples, Italy*
Marco Salvatore - *Naples, Italy*
Francesco Scolari - *Brescia, Italy*
Francesca Simonelli - *Naples, Italy*
Vicente E. Torres - *Rochester (MN), USA*
Francesco Trepiccione - *Naples, Italy*
Tivadar Tulassay - *Budapest, Hungary*
Robert Unwin - *London, UK*
Carsten Wagner - *Zurich, Switzerland*
David Warnock - *Birmingham (AL), USA*
Xueqing Yu - *Guangzhou, China*
Miriam Zacchia - *Naples, Italy*
Carmine Zoccali - *Reggio Calabria, Italy*

The kidney in genetic and rare diseases

Thursday, October 27th

11.00 a.m.-01.00 p.m.	Meeting with patients affected by Genetic Rare Diseases. Representative of political world, civil society and journalist will take part to the meeting
01.00-02.00 p.m.	Welcome Cocktail and Registration
02.00-02.30 p.m.	Opening Ceremony

Session I — Advanced Technologies

Chairmen: **G. Andria** *(Naples, Italy)*, **F. Salvatore** *(Naples, Italy)*

02.30-02.50 p.m.	Nephroplex for the detection of genomic variants using NGS **V. Nigro** *(Naples, Italy)*
02.50-03.10 p.m.	Integration of metabolomics and proteomics in exploring rare diseases **M. Ruoppolo** *(Naples, Italy)*
03.10-03.30 p.m.	Stem cell isolation from the urine of patients with rare diseases **P. Romagnani** *(Florence, Italy)*
03.30-03.45 p.m.	Discussion
03.45-04.15 p.m.	*Coffee Break*

Session II — Rare Disorders with Renal Phenotype

Chairmen: **A. Colao** *(Naples, Italy)*, **F. Simonelli** *(Naples, Italy)*

04.15-04.35 p.m.	Drug repositioning in cystinosis **F. Emma** *(Rome, Italy)*
04.35-04.55 p.m.	Medullary sponge kidney disease **G. Gambaro** *(Rome, Italy)*
04.55-05.15 p.m.	Genetics and pathogenesis of atypical hemolytic uremic syndrome **M. Noris** *(Bergamo, Italy)*
05.15-05.30 p.m.	Discussion

Key Note Lecture

Chairman: **C. Zoccali** *(Reggio Calabria, Italy)*

05.30-06.00 p.m.	Renal Tumor caused by TFEB overexpression **A. Ballabio** *(Naples, Italy)*
06.00-06.15 p.m.	Discussion

Friday, October 28th

Session III — Tubulopathies

Chairmen: **F. Mallamaci** *(Reggio Calabria, Italy)*, **G. Montini** *(Milan, Italy)*

08.30-08.50 a.m.	Fanconi Syndrome **R. Unwin** *(London, UK)*
08.50-09.10 a.m.	From rare to common kidney disorders: the case of Uromodulin **O. Devuyst** *(Zurich, Switzerland)*
09.10-09.30 a.m.	Rare Renal Diseases associated with gain or loss of function mutations in the Calcium Sensing Receptor **J. Geibel** *(New Heaven (CT), USA)*
09.30-09.50 a.m.	MicroRNAs in renal tubulopathies - a novel therapeutic target **S. Jelen** *(Ariano Irpino, Italy)*
09.50-10.10 a.m.	Discussion

Key Note Lecture

Chairman: **L. Gesualdo** *(Bari, Italy)*

10.10-10.40 a.m.	Retarding renal disease progression and the case of kidney self-repair **G. Remuzzi** *(Bergamo, Italy)*
10.40-10.55 a.m.	Discussion
10.55-11.25 a.m.	*Coffee Break*

Session IV — Acid-Base and Tubulopathies

Chairmen: **A. Dal Canton** *(Pavia, Italy)*, **P. Menè** *(Rome, Italy)*

11.25-11.45 a.m.	Acid-base imbalance due to inappropriate stimulation of Pendrin **K. Kawahara** *(Tokyo, Japan)*
11.45-12.05 a.m.	Mechanism of acidosis in Gordon Syndrome **D. Eladari** *(Paris, France)*
12.05-12.25 a.m.	Why R589H mutation of AE1 determines distal Renal Tubular Acidosis **F. Trepiccione** *(Naples, Italy)*
12.25-12.40 a.m.	Discussion
12.40 a.m.-02.00 p.m.	*Lunch* Poster Session

Friday, October 28th

Session V — Ciliopathies - Genetic Stone Diseases

Chairmen: **G. Grandaliano** *(Foggia, Italy)*, **G. Marinangeli** *(Giulianova, Italy)*

02.00-02.20 p.m.	The oral facial digital type I syndrome: a rare form of inherited renal cystic disease **B. Franco** *(Naples, Italy)*
02.20-02.40 p.m.	The Bardet Biedl Syndrome **M. Zacchia** *(Naples, Italy)*
02.40-03.00 p.m.	Rare causes of kidney stones **O. Moe** *(Dallas (TX), USA)*
03.00-03.20 p.m.	Cystinuria: new reflections based on genotyping **P. Jaeger** *(Geneva, Switzerland)*
03.20-03.40 p.m.	Discussion
03.40-04.10 p.m.	*Coffee Break*

Session VI — Polycistic Kidney Disease

Chairmen: **L. Murer** *(Padua, Italy)*, **F. Scolari** *(Brescia, Italy)*

04.10-04.30 p.m.	New signaling pathways and therapeutic targets in polycystic kidney disease **M.J. Caplan** *(New Heaven (CT), USA)*
04.30-04.50 p.m.	The role and mechanism of excessive activation of alternative complement pathway in ADPKD progression **M. Changlin** *(Shangai, China)*
04.50-05.10 p.m.	Long-acting somatostatin analogues for ADPKD **N. Perico** *(Bergamo, Italy)*
05.10-05.30 p.m.	Therapies for PKD targeting cyclic AMP **V.E. Torres** *(Rochester (MN), USA)*
05.30-05.50 p.m.	Discussion

Saturday, October 29th

Session VII — Glomerulopathies

Chairmen: **A. C. Ferreira** *(Lisbon, Portugal)*, **D. Roccatello** *(Turin, Italy)*

08.30-08.50 a.m.	From extreme phenotypes to the pathophysiology of auto-immune renal diseases **P. Ronco** *(Paris, France)*
08.50-09.10 a.m.	Glomerulopathy of genetic origin **X. Yu** *(Guangzhou, China)*
09.10-09.30 a.m.	Hereditary podocytopathies in adults: the next generation **O. Boyer** *(Paris, France)*
09.30-09.50 a.m.	Exploring the genetic basis of Nephrotic Syndrome **M. Gigante** *(Bari, Italy)*
09.50-10.10 a.m.	Discussion

Key Note Lecture

Chairman: **C. Ronco** *(Vicenza, Italy)*

10.10-10.40 a.m.	Potential of endocannabinoid system-based drugs and plant cannabinoids for the treatment of rare kidney disease **V. Di Marzo** *(Naples, Italy)*
10.40-10.55 a.m.	Discussion
10.55-11.25 a.m.	*Coffee break*

Saturday, October 29th

Session VIII Genes and Renal Fibrosis

Chairmen: **M. Arici** *(Ankara, Turkey)*, **S. Castellino** *(Catania, Italy)*

11.25-11.45 a.m.	Pro-fibrotic genes and selective modulation of their signal transduction in the development of renal fibrosis **T. Tulassay** *(Budapest, Hungary)*
11.45-12.05 a.m.	Renal fibrosis and rare genetic variants in IgA nephropathy **F. Pesce** *(Bari, Italy)*
12.05-12.25 a.m.	Genome-wide association studies in chronic kidney disease: can it help clinical approach **A. Pani** *(Cagliari, Italy)*
12.25-12.40 a.m.	Discussion
12.40 a.m.-2.00 p.m.	*Lunch* Poster Session

Session IX Divalent Ions Tubulopathies

Chairmen: **G. Garibotto** *(Genoa, Italy)*, **P. Messa** *(Milan, Italy)*

02.00-02.20 p.m.	Rare diseases of phosphate disorders **C. Wagner** *(Zurich, Switzerland)*
02.20-02.40 p.m.	Genetic of magnesium disorders **Q. Qian** *(Rochester (MN), USA)*
02.40-03.00 p.m.	Role of nuclear receptors (FXR and LXR) in kidney diseases **M. Levi** *(Denver (CO), USA)*
03.00-03.15 p.m.	Discussion

Saturday, October 29th

Session X — Genes and Blood Pressure

Chairmen: **G. Conte** *(Naples, Italy)*, **A. Perna** *(Naples, Italy)*

03.15-03.35 p.m. Fabry Disease: blood pressure, proteinuria and progression of nephropathy
D. Warnock *(Birmingham (AL), USA)*

03.35-03.55 p.m. The role of Na-Cl cotransporter in Gitelman's Syndrome and other diseases
G. Gamba *(Mexico City, Mexico)*

03.55-04.15 p.m. The importance of Na-Cl cotransporter in genetic hypertension
G. Capasso *(Naples, Italy)*

04.15-04.35 p.m. Discussion

Special Talk

Chairman: **S. Coppola** *(Caserta, Italy)*

04.35-05.05 p.m. Challenges in diagnosing and managing rare genetic diseases in the developing world
V. Jha *(New Dheli, India)*

05.05-05.35 p.m. CME Questionnaire and Closing Remarks

General Information

Conference Venue

The venue of the Congress will be the
Aula Magna
Congress Centre
Via Partenope, 36
Naples - Italy

Secretariat during the Conference

The Secretariat will be open at the following times:
Thursday, October 27th, 2016 from 9.00 a.m. to 6.30 p.m.
Friday, October 28th, 2016 from 8.00 a.m. to 6.30 p.m.
Saturday, October 29th, 2016 from 8.00 a.m. to 5.30 p.m.

Official language

The official language of the Congress will be English.
Please note that the simultaneous translation will not be provided.

Continuing Medical Education (CME)

Planning Congressi S.r.l. is a CME provider - identification code no. 38 - and has assigned to the event no. 9 credits for Physicians for the following disciplines: internal medicine, oncology, pediatrics, pharmacology, nephrology, primary care, genetics, ophthalmology, hematology, cardiology, urology, endocrinology, geriatrics, metabolic diseases.
The event is also dedicated to: Biologists, Midwives, Oculists, Chemists, Epidemiologists, Audiologists, Medical genetic laboratory Technicians, Neonatologists, Therapists of the neuro and psychomotor developmental.
Please be aware that for the acquisition of credits is mandatory to have attended the 100% of the whole duration of the congress and selected at least the 75% of correct answers of the CME questionnaire.

Registration

The Conference is free to attend. Please register on the website **http://kidneyingenetic2016.com**.

Technical facilities

Facilities will be available for computer presentations and overhead projections. A business centre with PC (Powerpoint for Windows) will be available for check and preview of presentations. It is essential that speakers take their CD or USB flash drive to the business centre at least one hour before the session starts.

The centre will be open at the following times:

Thursday, October 27th, 2016 from 9.00 a.m. to 6.30 p.m.
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Saturday, October 29th, 2016 from 8.00 a.m. to 5.30 p.m.

Lunches and coffee breaks

Lunches and coffee breaks will be free of charge and served at the congress venue.

Certificates of Attendance

Certificates of attendance will be issued at the registration desk following full attendance of the congress.

Poster Sessions

Posters will be available for viewing in the lunch hall during the breaks.
Meeting attendees will also have the opportunity to meet the abstract authors to discuss their research and ask questions during the presentation times listed.

Organizing Secretariat

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