

International Conference:

# The kidney in genetic and rare diseases

*Organized by*

Prof. Giovambattista Capasso

**II University of Naples**

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Fondazione Internazionale Menarini  
Symposia: 297

Naples October 27-29, 2016



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 shed 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*

Naples October 27-29, 2016



## President of the Conference

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Giovambattista Capasso - *Naples, Italy*

## Faculty

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Generoso Andria - *Naples, Italy*  
Mustafa Arici - *Ankara, Turkey*  
Andrea Ballabio - *Naples, Italy*  
Tullio Bongo - *Ariano Irpino, Italy*  
Michael Joseph Caplan - *New Haven (CT), USA*  
Santina Castellino - *Catania, Italy*  
Annamaria Colao - *Naples, Italy*  
Giuseppe Conte - *Naples, Italy*  
Salvatore Coppola - *Caserta, Italy*  
Antonio Dal Canton - *Pavia, Italy*  
Mario De Felice - *Naples, Italy*  
Natale G. De Santo - *Naples, Italy*  
Vincenzo Di Marzo - *Naples, Italy*  
Dominique Eladari - *St Denis de La Réunion, France*  
Francesco Emma - *Rome, Italy*  
Ana Carina Ferreira - *Lisbon, Portugal*  
Brunella Franco - *Naples, Italy*  
Giorgio Fuiano - *Naples, Italy*  
Gerardo Gamba - *Mexico City, Mexico*  
Giovanni Gambaro - *Rome, Italy*  
Giacomo Garibotto - *Genoa, Italy*  
John Geibel - *New Haven (CT), USA*  
Gregory Germino - *Bethesda (MD), USA*  
Loreto Gesualdo - *Bari, Italy*  
Maddalena Gigante - *Bari, Italy*  
Olivia Gillion Boyer - *Paris, France*  
Giuseppe Grandaliano - *Foggia, Italy*  
Philippe Jaeger - *Geneva, Switzerland*  
Katsumasa Kawahara - *Tokyo, Japan*  
Sanjib Kumar Sharma - *Dharan, Nepal*  
Moshe Levi - *Denver (CO), USA*  
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*  
Eric Olinger - *Zurich, Switzerland*  
Antonello Pani - *Cagliari, Italy*  
Norberto Perico - *Bergamo, Italy*  
Alessandra Perna - *Naples, Italy*  
Laura Perrone - *Naples, Italy*  
Francesco Pesce - *Bari, Italy*  
Federica Petrillo - *Ariano Irpino, Italy*  
Claudio Pisano - *Ariano Irpino, Italy*  
Gabriella Pocsafalvi - *Naples, 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*  
Domenico Russo - *Naples, Italy*  
Francesco Salvatore - *Naples, Italy*  
Marco Salvatore - *Naples, Italy*  
Franz Schaefer - *Heidelberg, Germany*  
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 Zaccchia - *Naples, Italy*  
Ortensio Zecchino - *Ariano Irpino, Italy*

## Thursday, October 27<sup>th</sup>

**11.00 a.m.-01.00 p.m.** Meeting with patients affected by Genetic and 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

*Chairpersons: **F. Salvatore** (Naples, Italy), **F. Simonelli** (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

*Chairpersons: **G. Andria** (Naples, Italy), **A. Colao** (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

*Chairperson: **M.J. Caplan** (New Heaven (CT), USA)*

**05.30-06.00 p.m.** Renal Cell Carcinoma due to TFEB overexpression  
**A. Ballabio** (Naples, Italy)

**06.00-06.15 p.m.** Discussion





## Friday, October 28<sup>th</sup>

### Session III Tubulopathies

*Chairpersons: **G. Fuiano** (Naples, 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  
**E. Olinger** (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.** Rare diseases of phosphate disorders  
**C. Wagner** (Zurich, Switzerland)
- 09.50-10.10 a.m.** Discussion

#### Key Note Lecture

*Chairperson: **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

*Chairpersons: **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** (St Denis de La Réunion, 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 28<sup>th</sup>

### Session V

### Ciliopathies - Genetic Stone Diseases

*Chairpersons: 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

### Polycystic Kidney Disease

*Chairpersons: 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.** Can lifestyle modifications alter the progression of PKD?  
**G. Germino** (Bethesda (MD), USA)
- 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

### Special Talk

*Chairperson: L. Perrone (Naples, Italy)*

- 05.50-06.15 p.m.** The translation of research insights to clinical practice - focusing on the ERN process  
**F. Schaefer** (Heidelberg, Germany)



## Saturday, October 29<sup>th</sup>

### Session VII

### Glomerulopathies

*Chairpersons: 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. Gillion 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

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

**10.10-10.40 a.m.** Potential of endocannabinoid system-based drugs and plant cannabinoids for the treatment of kidney disease

**V. Di Marzo** (Naples, Italy)

**10.40-10.55 a.m.** Discussion

**10.55-11.25 a.m.** *Coffee break*

## Saturday, October 29<sup>th</sup>

### Session VIII Genes and Renal Fibrosis

*Chairpersons: 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

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

- 02.00-02.20 p.m. MicroRNAs in renal tubulopathies - a novel therapeutic target  
**F. Petrillo** (Ariano Irpino, Italy)
- 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 29<sup>th</sup>

### Session X

### Genes and Blood Pressure

*Chairpersons: 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. Transporters and channels in genetic hypertension  
**G. Capasso** (Naples, Italy)
- 04.15-04.35 p.m. Discussion

### Special Talk

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

- 04.35-04.55 p.m. Prevention program for CKD in the developing world: challenges in diagnosing and managing rare genetic diseases  
**S.K. Sharma** (Dharan, Nepal)
- 04.55-05.05 p.m. Discussion
- 05.05-05.35 p.m. CME Questionnaire and Closing Remarks

## General Information

### Conference Venue

The venue of the Conference 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 27<sup>th</sup>, 2016 from 9.00 a.m. to 6.30 p.m.

Friday, October 28<sup>th</sup>, 2016 from 8.00 a.m. to 6.30 p.m.

Saturday, October 29<sup>th</sup>, 2016 from 8.00 a.m. to 5.30 p.m.

### Official language

The official language of the Conference 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.

Credits will be assigned also 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 Conference 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>.



Naples October 27-29, 2016

## Technical facilities

Facilities will be available for computer presentations. A Speakers' Ready Room with PC (Powerpoint for Windows) will be available for check and preview of presentations. It is essential that speakers deliver their presentation to the Speakers' Ready Room at least one hour before the session starts.

## The Speakers' Ready Room will be open at the following times:

Thursday, October 27<sup>th</sup>, 2016 from 9.00 a.m. to 6.30 p.m.

Friday, October 28<sup>th</sup>, 2016 from 8.00 a.m. to 6.30 p.m.

Saturday, October 29<sup>th</sup>, 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 Conference venue.

## Certificates of Attendance

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

## 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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### **PLANNING**

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# The kidney in genetic and rare diseases

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Leading  
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