International Conference:
The kidney in genetic and rare diseases

Organized by
Prof. Giovambattista Capasso
II University of Naples

Promoted by
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*
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 Gambardella - 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 - Geneve, Switzerland
Sabina Jelen - Ariano Irpino, Italy
Vivekanand Jha - New Delhi, 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 Scollarì - 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
Thursday, October 27th

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
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)*

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

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

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<tr>
<th>Time</th>
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<tbody>
<tr>
<td>10.10-10.40 a.m.</td>
<td>Retarding renal disease progression and the case of kidney self-repair</td>
<td>G. Remuzzi (Bergamo, Italy)</td>
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<tr>
<td>10.40-10.55 a.m.</td>
<td>Discussion</td>
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<td>10.55-11.25 a.m.</td>
<td>Coffee Break</td>
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**Session IV**  
*Acid-Base and Tubulopathies*

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

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

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<tr>
<th>Time</th>
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| 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)*

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</table>
| 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
### Session X  
**Genes and Blood Pressure**  
*Chairmen: G. Conte (Naples, Italy), A. Perna (Naples, Italy)*

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<tr>
<th>Time</th>
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<tbody>
<tr>
<td>03.15-03.35</td>
<td>Fabry Disease: blood pressure, proteinuria and progression of nephropathy</td>
<td>D. Warnock</td>
<td>Birmingham (AL), USA</td>
</tr>
<tr>
<td>03.35-03.55</td>
<td>The role of Na-Cl cotransporter in Gitelman’s Syndrome and other diseases</td>
<td>G. Gamba</td>
<td>Mexico City, Mexico</td>
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<tr>
<td>03.55-04.15</td>
<td>The importance of Na-Cl cotransporter in genetic hypertension</td>
<td>G. Capasso</td>
<td>Naples, Italy</td>
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<tr>
<td>04.15-04.35</td>
<td>Discussion</td>
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#### Special Talk  
*Chairman: S. Coppola (Caserta, Italy)*

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<tr>
<td>04.35-04.55</td>
<td>Challenges in diagnosing and managing rare genetic diseases in the developing world</td>
<td>V. Jha</td>
<td>New Delhi, India</td>
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<tr>
<td>04.55-05.05</td>
<td>Discussion</td>
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<tr>
<td>05.05-05.35</td>
<td>CME Questionnaire and Closing Remarks</td>
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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.
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 congress and selected at least the 75% of correct answers of the CME
questionnaire.

Registration
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.
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.

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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