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6th Symposium on Nephrolithiasis
of the Fondazione Menarini
GENETICS OF NEPHROLITHIASIS
and
**Nephrolithiasis for Nephrologists:
an introductory course**

8th-11th May 2024 - HOTEL EXE LEV Vošnjakova ulica 1, 1000 Ljubljana, Slovenia

The most severe forms of nephrolithiasis are Mendelian disorders. Although rare, these forms should be diagnosed because they may have specific treatments. Their recognition is key to personalized/precision medicine. It is the case of type I primary hyperoxaluria, a dramatic condition that has now a treatment.

Many of the monogenic forms of nephrolithiasis are associated with nephrocalcinosis. This is an almost neglected condition of which even the definition is unclear. To define what nephrocalcinosis is, the meeting will start with the presentation of the preliminary results of a Consensus Document written by a group of experts that join the competence in bone and mineral abnormalities of CKD (the working group of ERA on CKD-MBD) and the bone and mineral abnormalities of nephrolithiasis (the Faculty of the 6th Symposium).

We are facing a sort of crisis in treating patients with nephrolithiasis. On one side, the terrific technological advancement that has brought in the surgical theatre tools for truly mini-invasive treatments of stones, absorbing almost totally the attention of urologists who have less and less time for stone metaphylaxis; on the other, very few nephrologists know how to do stone metaphylaxis because the easy non-invasive removal of stones is perceived as sufficient. However, behind nephrolithiasis, severe conditions may be present, including the genetic forms that frequently lead to CKD and ESKD, among many others. Their diagnosis is crucial for giving specific treatments. However, even in the idiopathic cases of nephrolithiasis, the quality of life of the patient may be compromised because of pain, hospital admissions, urological treatments, days lost at work, etc. These patients, too, need efficacious treatments. This is why we, in cooperation with the Slovenian Society of Nephrology, have dedicated the last part of the Symposium to an introductory course on Nephrolithiasis for nephrologists. We hope that this will help to spread the knowledge and the skills necessary to nephrologists for treating patients with nephrolithiasis, to give them better prognosis and quality of life.

Emanuele Croppi

Giovanni Gambaro

Pietro Manuel Ferraro

Andrej Škoberne

UNDER THE AUSPICES OF



PRESIDENTS

Emanuele Croppi (Florence, IT)

Giovanni Gambaro (Verona, IT)

Pietro Manuel Ferraro (Rome, IT)

Andrej Škoberne (Ljubljana, SL)

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GENETICS OF NEPHROLITHIASIS

and Nephrolithiasis for Nephrologists: an introductory course

PROGRAMME

THURSDAY, 9th MAY

WEDNESDAY, 8th MAY 2024 - AFTERNOON

05.00 p.m. - 07.00 p.m. Faculty meeting

THURSDAY, 9th MAY 2024 - MORNING

08.00 a.m. - 10.00 a.m. Consensus document on nephrocalcinosis
(**P. M. Ferraro** and the Faculty, by invitation only)

10.00 a.m. - 10.20 a.m. *Coffee Break*

10.20 a.m. - 10.30 a.m. Welcome and Presentation of the Symposium
(Presidents of the Meeting and Fondazione Menarini)

SESSION I - GENETICS OF METABOLIC DISORDERS CAUSING NEPHROLITHIASIS

Chairpersons: **F. Emma** (Rome, IT), **S. Moochhala** (London, UK)

10.30 a.m. - 11.00 a.m. Primary hyperoxalurias: Clinical phenotypes and treatment
J. Groothoff (Amsterdam, NL)

11.00 a.m. - 11.30 a.m. Primary hyperoxalurias: Curation of genetic variants to assist in diagnostic accuracy and estimation of the true prevalence of primary hyperoxaluria
G. Rumsby (London, UK)

11.30 a.m. - 12.00 a.m. CYP24A1 gene mutations and polymorphisms in renal stone formers
K. P. Schlingmann (Munster, DE)

12.00 p.m. - 12.30 p.m. Diamonds in the kidney: crystal nephropathies
F. Knauf (Berlin, DE)

12.30 p.m. - 02.00 p.m. *Light Lunch*

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THURSDAY, 9th MAY

THURSDAY, 9th MAY 2024 - AFTERNOON

SESSION II - GENETICS OF RENAL DISORDERS CAUSING NEPHROLITHIASIS

Chairpersons: **G. Vezzoli** (Milan, IT),
N. Maalouf (Dallas, USA)

- 02.00 p.m. - 02.30 p.m. Impaired phosphate tubular handling and genetic nephrolithiasis
C. Wagner (Zurich, CH)
- 02.30 p.m. - 03.00 p.m. ABC(C) tale of pyrophosphate in nephrolithiasis: from the rare to the general?
E. Letavernier (Paris, FR)
- 03.00 p.m. - 03.30 p.m. The “inflamed” Cystinuria
M. Courbebaïsse (Paris, FR)

SESSION III - THE CONTRIBUTION OF GENETICS TO DIAGNOSIS AND PATHOGENESIS - PART I

Chairpersons: **M. Marangella** (Turin, IT),
R. Torra (Barcelona, ES)

- 03.30 p.m. - 04.00 p.m. Three W in Genetic testing in renal stone formers (When, in Whom, Why)
J. Halbritter (Berlin, DE)
- 04.00 p.m. - 04.30 p.m. The complexity of the genetic risk of mineral and stone disorders: dissecting it by GWAS
S. Howles (Oxford, UK)
- 04.30 p.m. - 05.00 p.m. Genetics of electrolyte control: insights from GWAS and PheWAS
M. Treccani (Verona, IT)
- 05.00 p.m. - 05.30 p.m. *Coffee Break*

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PROGRAMME

THURSDAY, 9th MAY

**THE CONTRIBUTION OF GENETICS TO DIAGNOSIS
AND PATHOGENESIS - PART II**

Chairpersons: **J. Halbritter** (Berlin, DE),
J. Williams (Indianapolis, USA)

- 05.30 p.m. - 06.00 p.m. Rare mendelian diseases and nephrolithiasis, variable phenotypes, digenic disorders: how much rare is rare?
J. Lieske (Rochester, USA)
- 06.00 p.m. - 06.30 p.m. Epigenetics, metabolomic and nephrolithiasis
P. M. Ferraro (Rome, IT)
- 06.30 p.m. - 07.00 p.m. Big data, the exposome and genome: how artificial intelligence can help research on stones?
D. Goldfarb (New York, USA)

GENETICS OF NEPHROLITHIASIS

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PROGRAMME

FRIDAY, 10th MAY

FRIDAY, 10th MAY 2024 - MORNING

SESSION IV - NEPHROLITHIASIS FOR NEPHROLOGISTS: AN INTRODUCTORY COURSE Jointly with the **Slovenian Society of Nephrology**

Chairpersons: **E. Letavernier** (Paris, FR),
G. Curhan (Boston, USA)

- 08.30 a.m. - 09.00 a.m. Metabolic and infective factors in nephrolithiasis: epidemiological and pathogenetic evidences
A. Trinchieri (Milan, IT)
- 09.00 a.m. - 09.30 a.m. Metabolic disorders in nephrolithiasis
what can we find in the urine and how do we interpret the findings?
P. M. Ferraro (Rome, IT)
- 09.30 a.m. - 10.00 a.m. Nutritional treatment in the stone patient
A. Cupisti (Pisa, IT)
- 10.00 a.m. - 10.30 a.m. Treatment after metabolic evaluation
D. Fuster (Bern, CH)
- 10.30 a.m. - 10.45 a.m. *Coffee Break*
- 10.45 a.m. - 11.15 a.m. Treating a kidney stone patient when metabolic evaluation is not available:
all the good things a nephrologist can and should do
D. Goldfarb (New York, USA)
- 11.15 a.m. - 11.45 a.m. Chronic kidney disease, cardiovascular disease and bone disease in nephrolithiasis:
looking beyond the stones
A. Škoberne (Ljubljana, SL)

GENETICS OF NEPHROLITHIASIS
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PROGRAMME

FRIDAY, 10th MAY

Chairpersons: **C. Vitale** (Turin, IT),
O. Bonny (Fribourg, CH)

11.45 a.m. - 01.40 p.m. Kidney stone patient in the nephrology outpatient clinic:
clinical cases

O. Bonny (Fribourg, CH) , **G. Capolongo** (Naples,IT),
C. Vitale (Turin, IT)

01.40 p.m. - 02.40 p.m. *Light Lunch*

Chairpersons: **G. Gambaro** (Verona, IT),
A. Škoberne (Ljubljana, SL)

02.40 p.m. - 03.30 p.m. Kidney stone patient in the nephrology outpatient clinic:
clinical cases (continuation)

S. Moochhala (London, UK)

03.30 p.m. - 04.00 p.m. Final messages and conclusions

SATURDAY, 11th MAY 2024 - MORNING

09.00 a.m. - 11.00 a.m. Faculty meeting

GENETICS OF NEPHROLITHIASIS
and Nephrolithiasis for Nephrologists: an introductory course

FACULTY

Olivier Bonny

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Fribourg, CH

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Naples, IT

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Adamasco Cupisti

Full Professor of Nephrology
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Gary Curhan

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GENETICS OF NEPHROLITHIASIS

and Nephrolithiasis for Nephrologists: an introductory course

FACULTY

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Jaap Groothoff

MD PhD | Pediatric Nephrologist
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GENETICS OF NEPHROLITHIASIS
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FACULTY

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GENETICS OF NEPHROLITHIASIS
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FACULTY

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Roser Torra Inherited Kidney Diseases
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Corrado Vitale Director SC Nephrology and Dialysis
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Turin, Italy

GENETICS OF NEPHROLITHIASIS
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FACULTY

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James Williams

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Indianapolis, USA

GENETICS OF NEPHROLITHIASIS and Nephrolithiasis for Nephrologists: an introductory course

REGISTRATION - HYBRID EVENT

VIA WEBSITE AND/OR APP - HYBRID EVENT

Subscription to the Reserved Area and to the Event of your interest is required.
REGISTRATION IS FREE.

RESIDENTIAL EVENT

Via website www.en.fondazione-menarini.it/Home

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3) Fill in all the mandatory fields and complete the registration, click on **REGISTER** (only once).

Wait for the confirmation page to appear, do not click on anything or leave the page.

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GENETICS OF NEPHROLITHIASIS and Nephrolithiasis for Nephrologists: an introductory course

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REGISTRATION - FONDAZIONE APP



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GENERAL INFORMATION

VENUE

HOTEL EXE LEV

Vošnjakova ulica 1, 1000 Ljubljana, Slovenia

OFFICIAL LANGUAGE

The official language of the meeting is English. Simultaneous translation will not be provided.

REGISTRATION HOURS FOR RESIDENTIAL EVENT

Attendance at the meeting is free.

For more information and to register please visit the following website:

www.en.fondazione-menarini.it/Home

Thursday, 9th May 2024: 07.00 a.m. - 07.00 p.m.

Friday, 10th May 2024: 07.30 a.m. - 04.00 p.m.

Please note that times are expressed in Local time

FACULTY SLIDE CENTRE OPENING HOURS

The center will be open at the following times:

Thursday, 9th May 2024: 07.00 a.m. - 07.00 p.m.

Friday, 10th May 2024: 07.30 a.m. - 04.00 p.m.

Please note that times are expressed in Local time

TECHNICAL FACILITIES

Facilities will be available for computer presentations and overhead projections.

A Slide Center with PCs (Windows and Macintosh operating systems) will be available for

Speakers to preview and finalize their presentations. Speakers are kindly requested to submit their presentations to the Slide Center technicians at least one hour before their talks.

LUNCH AND COFFEE BREAKS

Lunch and coffee breaks will be served in the meeting area.

ATTENDANCE CERTIFICATE

The attendance certificate will be provided via email after the meeting, upon request.



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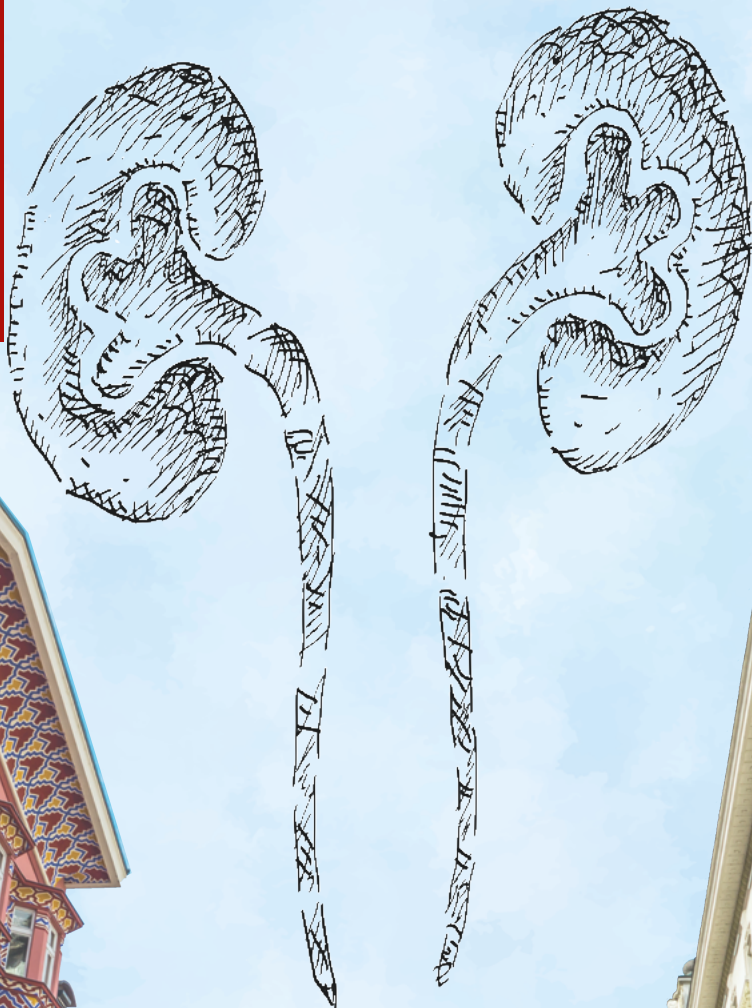
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