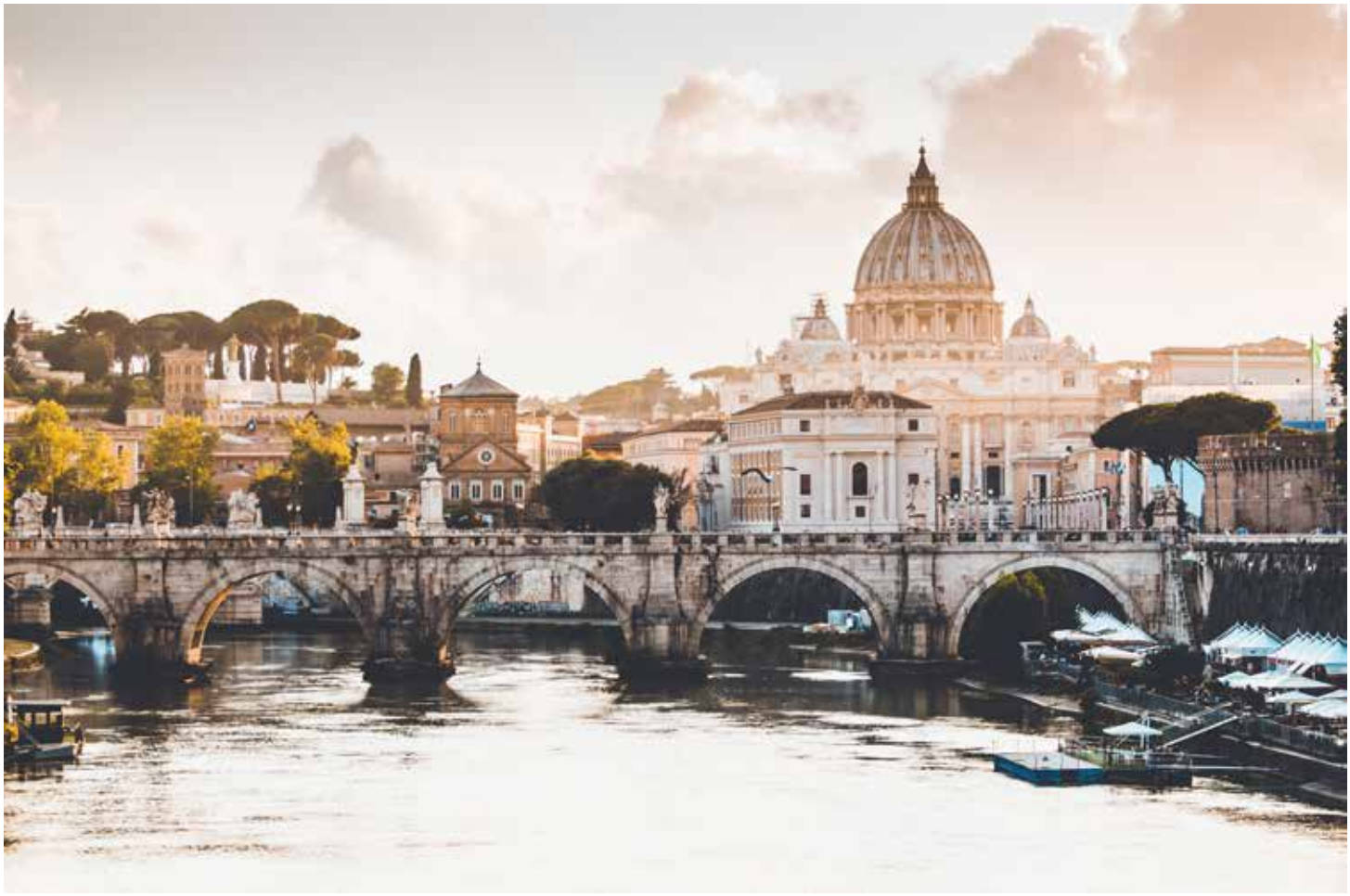

THIRD INTERNATIONAL HOMOCYSTINURIAS

# PATIENT-EXPERT MEETING

28 FEBRUARY - 1 MARCH 2019 IN ROME, ITALY

## COLLABORATING PATIENT ORGANIZATIONS AND CONSORTIA

HCU Network Australia

EHOD
European Network and Registry for Homocystinurias and Methylation Defects

cblC onlus

cblC Brasil
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HCU Network America

ASSOCIAZIONE ONLUS "LA VITA È UN DONO"

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for rare or low prevalence complex diseases
Network Hereditary Metabolic Disorders

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# THIRD INTERNATIONAL HOMOCYSTINURIAS PATIENT-EXPERT MEETING

## THURSDAY 28 FEBRUARY & FRIDAY 1 MARCH 2019

**NH Collection Roma Centro**
Via dei Gracchi, 324 00192 Rome - Italy

### REGISTRATION:

Located in the foyer area of the Sala Aracoeli & Campidoglio rooms
Open from 8.00am both Thursday & Friday

### CONFERENCE:

THURSDAY 8.30am to approx. 4.45pm
FRIDAY 8.30am to approx. 3.30pm

## CONTENTS

# GETTING THERE

## HOW TO ARRIVE IN ROME

**Leonardo da Vinci (Fiumicino) Airport (FCO)**
Leonardo da Vinci Airport is the main airport serving Rome. The airport is located 29km from the city centre of Rome. There are several bus, taxi and transfer companies who provide airport shuttle services.

**Ciampino Airport (CIA)**
If you are travelling from or to another area within Italy, Ciampino Airport is the airport serving domestic and some international flights. The airport is located 15km from the city center of Rome. Bus, taxi and transfer companies provide airport shuttle services.

**Stazione Termini**
Stazione Termini is the main train station in Rome. Airport buses and trains, as well as international trains will arrive at Stazione Termini. Here you can also get a taxi and bus to take you on to your destination.

# ABOUT ROME

As Italy's capital, Rome is one of the countries most popular tourist destinations.

Some suggested places to visit in Rome:

- Colosseum
- St. Peter's Basilica
- Pantheon
- Sistine Chapel
- Trevi Fountain
- Vatican City

# ACCOMMODATION

For participants who have reserved accommodation at NH Collection Roma Centro check in time is from 3pm and check out time is 12 noon. HCU Network Australia has secured a room block at the NH Collection Roma Centro. The hotel accomodations are processed through the hotel directly using the booking link for participants here. Please click here to be taken to the Roma Centro booking link to book your hotel room and to read the room cancellation policy. The room block will close on 31 January 2019.

The hotel offers

- Café-Bar open breakfast, lunch and dinner
- Restaurant – open lunch and dinner
- Gym
- Free Wi-Fi throughout the hotel
- Concierge services
- Currency exchange
- Dry cleaning service
- Express laundry service
- 24 hour front desk
- Air conditioning
- Multilingual staff

For more information on rooms please visit the hotel website directly.

# MEALS

Breakfast is available at the Café for Earlybird Breakfast 5am to 6.45am Monday to Sunday and Breakfast 7am to 10am Monday to Friday and Saturday & Sunday 7am to 11am.

Coffee breaks and lunch will be catered for as part of the Conference on Thursday & Friday.

Dinner will be at your own cost and arrangement.

## DIETARY REQUIREMENTS

Dietary requirements advised when you booked or via email to info@hcunetworkaustralia.org.au by **1 February 2019** will be catered for.

# LOCAL INFO

**Language:** Italian is the official and most widely spoken language in Rome. As Rome is a large tourist city, many people also speak English.

**Time Zone:** Rome follows the Central European Time (CET) (UTC/GMT + 1 hours)

**Weather:** With the end of winter for Rome in February and the beginning of Spring in March, the average maximum temperature is 15 C and the average minimum is 5 C.

**Dress code:** The dress code during the Conference is smart casual attire.

**Electricity:** You will require a travel adapter for your electrical goods.

**Telecommunications:** The Country Code for Italy is +39 and the area code for Rome is 06.

**Currency:** The Italian monetary unit is the Euro (EUR).

**Credit Cards:** Visa, MasterCard, Diners and American Express are accepted in most hotels, restaurants and shops.

**Visas:** Australian citizens planning on staying within Italy or other EU member countries for less than 90 days are not required a visa.

**Taxis:** When taking a taxi in Rome make sure the meter reads Tariffa 1. Sometimes, taxi drivers will put the meter on Tariffa 2 for unsuspecting tourists, which charges the passenger at a much higher rate. Tariffa 2 must only be applied when exiting Rome's highway, Grande Raccordo Annulare.

**Tipping:** Tips are optional and not expected in Italy. However, tipping may be appropriate where service is exceptional.

**Emergency Numbers:**

Police (Carabineri) – 112

General Emergency – 113

Fire Brigade – 115

Car Breakdown Assistance – 116

Ambulance / Medical Emergencies – 118

# QUESTIONS

Prior to the event please contact HCU Network Australia at info@hcunetworkaustralia.org.au

On the day, the Registration desk will be located in the foyer area of the Sala Aracoeli & Campidoglio rooms and will be open from 8.00am.

# SPONSORS

Once again we would like to thank our 2019 Conference partners.

Please see a comprehensive list of sponsors and their bios on the following pages.

# THURSDAY 28 FEBRUARY

| | | |
|---|---|---|
| 8.00 | Registration open | |
| 08.30-10.30 | **SESSION 1: State of the art lectures** | Chair: Ida SCHWARTZ |
| | Opening address: | Carlo DIONISI-VICI<br>Tara MORRISON |
| | Metabolism | |
| 8.30 – 8.50 | Sulfur Amino Acids | Viktor KOZICH |
| 8.50 – 9.10 | Folate and B12 | Henk BLOM |
| | Clinical Presentations | |
| 9.10 – 9.30 | CBS deficiency: Overview | Sufin YAP |
| 9.30 – 9.40 | CBS deficiency: A parents' perspective | Tara MORRISON |
| 9.40 – 10.00 | CblC and MTHFR: Overview | Carlo DIONISI-VICI |
| 10.00 – 10.10 | CblC: an adults' perspective | Francesca RESTUCCIA |
| 10.10 | COFFEE BREAK | |
| 10.30-11.10 | **SESSION 2: Novel Developments in Remethylation Defects** | Chair: Henk BLOM |
| 10.30 -10.50 | A transgenerational MMACHC epimutation illustrates a new mechanism of inborn errors of metabolism named epi-cblC | Jean-Louise GUEANT |
| 10.50 -11.10 | Structural basis of severe 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency | Matthias BAUMGARTNER |
| 11.10 – 13.10 | **SESSION 3: Newborn screening** | Chair: Alberto BURLINA |
| 11.10 – 11.40 | EHOD Recommendations on newborn screening for homocystinurias versus current practices | Martina HUEMER |
| 11.40 – 12.00 | Technical aspects of NBS in Homocystinurias | Giancarlo LA MARCA |
| 12.00 – 12.20 | Clinical management of a positive NBS for Homocystinurias | Alberto BURLINA |
| 12.20 – 12.50 | The outcomes in classical homocystinuria patients detected by newborn screening | Tawfeg BEN-OMRAN |
| 12.50 – 13.10 | Novel Developments in Remethylation Defects and Newborn screening: Q & A | Session speakers |
| 13.10 | LUNCH | |
| 14.00 – 15.30 | **SESSION 4: Interactive Panel** | Chair: Kimberly CHAPMAN |
| 14.00 – 14.45 | CBS deficiency | Viktor KOZICH<br>Tawfeg BEN-OMRAN<br>Sufin YAP |
| 14.45 – 15.30 | Remethylation disorders | Martina HUEMER<br>Carlo DIONISI-VICI<br>Matthias BAUMGARTNER<br>Irini MANOLI |
| 15.30 | COFFEE BREAK | |
| | **SESSION 5: New Developments** | Chair: Viktor KOZICH |
| 15.50 – 16.10 | Microbiome in CBS deficiency | Ida SCHWARTZ |
| 16.10 – 16.30 | Arg to Cys mutation in CBS deficiency | Henk BLOM |
| 16.30 – 16.45 | Closing comments: Day 1 | Henk BLOM<br>Ida SCHWARTZ |
| 17.00 – 18.00 | NETWORKING CANAPES & DRINKS | |

# FRIDAY 1 MARCH

| | | |
|---|---|---|
| 8.00 | Registration open | |
| 08.30-10.30 | **SESSION 6: Long term outcomes** | Chair: Carlo DIONISI-VICI |
| 8.30 – 9.00 | E-HOD Registry results: Remethylation disorders | Martina HUEMER |
| | E-HOD Registry results: CBS deficiency | Viktor KOZICH |
| 9.00 – 9.20 | The Irish Experience | Gregory PASTORES |
| 9.20 – 9.40 | Observational study of the natural history of patients with classical homocystinuria on current therapy | Marcia SELLOS-MOURA |
| 9.40 – 11.00 | **SESSION 7: Recent advances in novel treatments** | Chair: TBC |
| 9.40 – 10.00 | Enzyme replacement therapy for classical homocystinuria | Tomas MAJTAN |
| 10.00 – 10.20 | Impact of EPI743 on visual function in cblC defect | Diego MARTINELLI |
| 10.20 – 10.40 | Progress in CblC treatment | Irini MANOLI |
| 10.40 – 10.50 | Treatment of cobalamin metabolic disorders with oh-cobalamin through an “in situ” subcutaneous permanent catheter: reaching the goal. | Andrea BORDUGO |
| 10.50 – 11.00 | Long term outcomes and novel treatments: Q & A | Session Speakers |
| 11.00 | COFFEE BREAK | |
| 11.20 – 13.00 | **SESSION 8: Living with homocystinuria** | Chair: Ida SCHWARTZ |
| 11.20 – 12.00 | Dietary practices in pyridoxine non-responsive homocystinuria and the impact of E-HOD guidelines | Marjorie DIXON<br>Alexandra JUNG |
| 12.00 – 12.15 | RMD calculator& platform- tools supporting HCU diet calculation and communication between HCP & patients | Iwona BARTLOMIEJCZYK |
| 12.15 – 12.25 | Classical Homocystinuria in Qatar: Nutritional advances and challenges | Reem AL-SAADI |
| 12.25 – 12.40 | The challenges of parenting a child with phenylketonuria (PKU): Parents’ lived experiences | Anja WITTKOWSKI |
| 12.40 – 12.45 | Growing up with classical homocystinuria: a child’s perspective. | Owen AUSTIN |
| 12.45 – 13.00 | Living with homocystinuria: Q & A | Session speakers |
| 13.00 | LUNCH | |
| 14.00 – 15.30 | **SESSION 9: Patient Organisations** | Chair: Tara MORRISON |
| 14.00 – 14.20 | Global Research Map and Grants Process for HCU | Margie MCGLYNN |
| 14.20 – 14.40 | Homocystinuria Patient and Caregiver Survey: Experiences of Diagnosis and Patient Satisfaction | Tara MORRISON |
| 14.40 – 15.00 | Cblc onlus, from the beginning until now | Rossella BRINDISI |
| 15.00 – 15.20 | cblC Brazil - News and Progress | Luana BRITO |
| 15.20 – 15.30 | Closing comments: Day 2 | Henk BLOM<br>Ida SCHWARTZ |
| | Acknowledgements | Carlo DIONISI-VICI<br>Tara MORRISON |
| 15.30 | CLOSE | |

# SILVER SPONSORS


Orphan Europe, part of the Recordati Group, is a pharmaceutical company aiming at providing treatment for patients with unmet medical needs suffering from rare diseases. Since 1990, Orphan Europe is established as one of the most active players in the field of rare diseases.

Orphan Europe is committed to the improvement of knowledge about rare diseases among healthcare professionals. The best example is the support of Recordati Rare Diseases Foundation that provides unique, independent and high-level education on rare diseases.

Orphan Europe shares the conviction that every single patient has the right to the best possible treatment.

For more information, please visit www.orphan-europe.com


Orphan Technologies is dedicated to developing novel therapies to dramatically improve the lives of patients suffering from rare disorders. OT-58, our lead drug development candidate, has been optimized as an enzyme replacement therapy for classical homocystinuria, a genetic disease characterized by debilitating cardiovascular, skeletal, neurologic, and ophthalmologic complications. OT-58 is designed to reduce homocysteine levels via a targeted mechanism of action and may have therapeutic applications in other diseases. For more information, please visit www.orphantechnologies.com.

OT-58, Orphan Technologies' lead drug candidate, is a modified recombinant enzyme replacement therapy in development as an enzyme replacement therapy for patients suffering from the rare disease classical homocystinuria. Classical homocystinuria is a genetic metabolic disease caused by a deficiency in the CBS enzyme leading to elevated levels of the amino acid homocysteine. OT-58 has consistently demonstrated significant reductions in homocysteine levels across multiple models of homocystinuria and has the potential to improve metabolic control, reduce or remove dietary restrictions, and positively impact clinical outcomes. OT-58 is anticipated to enter clinical evaluation in 2018 and has been granted Orphan Status by the US Food and Drug Administration and EMA. In addition, based on its mechanism of action, OT-58 has therapeutic potential in other diseases.

For more information, please visit www.orphantechnologies.com

Nutricia pioneers nutritional solutions that help people live longer, more joyful and healthier lives.

Building on more than a century of nutritional research and innovation, Nutricia continues to transform lives through the power of nutrition.

Nutricia's science-based products and services support healthy growth and development during the first 1000 days. Nutricia also helps to address some of the world's biggest health challenges including conditions in early life such as pre-term birth, faltering growth, food allergy and rare metabolic diseases, as well as age-related conditions and chronic disease, such as frailty, cancer, stroke and early Alzheimer's disease.

As part of Danone, Nutricia fully embraces the company's "One Planet. One Health" vision reflecting that the health of people and the health of the planet are interconnected and therefore seeks to protect and nourish both.

For more information visit www.nutricia.com

## BRONZE SPONSORS

Founded in Genoa in 1915 by Edoardo Maragliano, the man who discovered the first tuberculosis vaccine, PIAM is an Italian pharmaceutical company that has developed its core business in specialist and ethically significant therapeutic areas with prescription drugs.

Over time it has gained particular expertise in hereditary metabolic diseases. In the 70s, PIAM pioneered a specialist nutritional line to support its pharmaceutical products. It is able to offer a complete response for patients with disorders requiring particular nutritional therapy, throughout every phase of their lives.

For more information, please visit www.piamfarmaceutici.com/en/

BioElectron is a platform biotechnology company that is using its expertise in redox chemistry to develop first-in-class therapeutics for CNS and non-CNS diseases characterized by redox defects. BioElectron's lead clinical compound—EPI-743—is in late clinical stage development for mitochondrial disease and related orphan disorders with shared biochemistry, including cobalamin C defect. In addition, BioElectron has a rich pipeline of platform-derived compounds in various stages of development from preclinical to phase 2.

For more information, please visit www.bioelectron.com

# BRONZE SPONSORS (CONT.)

Vitaflo are at the forefront of developing innovative specialised clinical nutrition products for Metabolic Disorders such as Homocystinuria and other specific conditions. Our aim is to create products that combine the best of cutting-edge research with the lifestyle demands of modern living, ensuring the most acceptable products are available for the patient. By constantly evolving to meet patient needs, Vitaflo will continue to develop products which offer patients choice and help support them in complying with restrictive therapeutic diets. For more information, please visit www.vitafloweb.com

metaX, works with prestigious universities and special hospitals, research laboratories and competent business partners. metaX offers dietetic Foods for Special Medical Purposes: Protein Supplements, powdered and liquid, a wide range of Low Protein Products for patients with PKU, HCU and many other rare disorders of metabolism, Supplements for Food Fortification, a Micronutrient Supplement System and products for the dietetic management of Chronic Renal Failure.

metaX services include brochures, recipes, cooking classes, holiday activities for patients and medical symposia for professionals.For more information visit www.metax.org

# SOCIETY SPONSORS

The aim of the Society of the Study of Inborn Errors of Metabolism is to foster the study of inherited metabolic disorders and related topics. The Society, founded in 1963, exists to promote the exchange of ideas between professional workers in different disciplines who are interested in inherited metabolic disease. Pursuing this aim by arranging scientific meetings, publications and in other ways considered appropriate by the Council. The Council is also supported by advisory council members who provide advice and meet with the Council at the annual symposium.

For more information, please visit www.ssiem.org

# SUPPORTERS

Cambrooke Therapeutics (expansion of Cambrooke Foods) was founded in 2000 by Lynn and David Paolella, the parents of two children diagnosed with a rare disease called phenylketonuria (PKU). PKU is one of the few genetic diseases, which is managed almost entirely with nutritional intervention. The Paolellas' goal in forming Cambrooke was simple – to develop improved nutritional therapeutic options for those with serious medical disorders.

For more information, please visit www.cambrooke.com