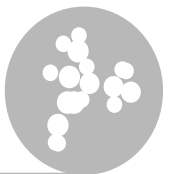




THIRD INTERNATIONAL HOMOCYSTINURIAS
PATIENT-EXPERT MEETING
28 FEBRUARY - 1 MARCH 2019 IN ROME, ITALY



COLLABORATING PATIENT ORGANIZATIONS AND CONSORTIA



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



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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 accommodations 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 Anulare.

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

Emergency Numbers:

Police (Carabinieri) - 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 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 Tawfeg BEN-OMRAN Sufin YAP
14.45 – 15.30	Remethylation disorders	Martina HUEMER Carlo DIONISI-VICI Matthias BAUMGARTNER Irina 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 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 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-SAAD
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 Ida SCHWARTZ
	Acknowledgements	Carlo DIONISI-VICI Tara MORRISON
15.30	CLOSE	



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.)



Vitaflor 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, Vitaflor 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 Paoella, 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 Paoellas' 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