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Supporters



11th International Workshop on Alkaptonuria (AKU)

Siena

30th January 2019

University of Siena

Dept. Biotechnology, Chemistry and Pharmacy

via Aldo Moro 2
room 14



Programme

9:00 Registration

9:30 Welcome message by

Prof. Francesco Frati, Rector

Prof. Annalisa Santucci, Head of Dept. Biotechnology, Chemistry and Pharmacy – Dept. of Excellence 2018-2022

Dr. Valtere Giovannini, General Director of Siena University Hospital, Regional Center for Precision Medicine

Prof. Antonio Federico, HCP Representative for MetabERN, Corporate and Regional Coordinator for Rare Diseases, Siena University Hospital

Dr. Cristina Tinti, Incubation Manager Fondazione TLS

Session 1: AKU – Italian medical framework (Chair: Prof. Frediani)

10:00 Dr. Marco Bardelli (Siena University Hospital, Italy) DTA (Percorso Diagnostico Terapeutico Assistenziale for AKU) Guidelines for the diagnosis and clinical management of AKU

10:15 Dr. Dario Gambera (Rugani Hospital, Siena, Italy) Orthopaedic surgical interventions in AKU patients

10:30 Dr. Giuseppe Greco (Rugani Hospital, Siena, Italy) Morphological studies on ex-vivo cultured AKU human anterior lens epithelial cells

10:45 Coffee break

Session 2: AKU – International medical framework (Chair: Prof. Ranganath)

11:15 Dr. Wendy Introne (NIH, USA) Alkaptonuria in the United States

12:00 Ms. Hollie Washington (RLBUH Liverpool, UK) UK National Alkaptonuria Centre and logistics

12:15 Dr. Jean-Baptiste Arnoux & Dr. Kim-Hanh Le Quan Sang (Necker Hospital, France) Different experiences about Nitisinone treatment for AKU in French Metabolite Reference Center

12:30 Dr. Mariusz Kujawa (Medical University of Gdansk, Poland) Care for paediatric patients with alkaptonuria

12:45 Dr. Isaac Jebaraj (Christian Medical College, Vellore, India) Alkaptonuria in India: an update

13:00 Prof. Maurizio Scarpa, Prof. Sylvia Sestini (MetabERN) MetabERN - European Reference Networks for hereditary metabolic disorders

13:15 Light lunch

Session 3: AKU imaging (Chair: Prof. Gallagher)

14:15 Dr. Leah Taylor (RLBUH Liverpool, UK) Assessment of AKU using quantitative image analysis

14:30 Prof. Jim Gallagher (University of Liverpool, UK) Joint destruction in AKU

14:45 Prof. Gabor Barton (Liverpool John Moore University, UK) Gait analysis in AKU

Session 4: Novel insights on AKU molecular mechanisms and therapeutics (Chair: Prof. Santucci)

15:00 Dr. Andrea Zatkova (Slovak Academy of Sciences, Slovakia) What does SONIA2 tell us about genetics of AKU

15:15 Dr. Anna Milan (RLBUH Liverpool, UK) The metabolic consequences of Nitisinone

15:30 Dr. Daniela Braconi (University of Siena, Italy) Molecular mechanisms of AKU physiopathology

15:45 Dr. Elena Petricci (University of Siena, Italy) Novel insights on HGA chemical transformation and Rational drug design and synthesis of novel HPPD inhibitors

16:00 Dr. Alfonso Trezza (University of Siena, Italy) An advanced Molecular Dynamics study to discover novel inhibitors of Human 4-hydroxyphenylpyruvate dioxygenase

16:15 Dr. Andrea Bernini (University of Siena, Italy) In silico studies of refolding chaperones for HGD

Session 5: Concluding remarks & future perspectives (Chairs: Prof. Santucci and Prof. Gallagher)

16:30 Dr. Vittoria Cicaloni (University of Siena, Italy) An AKU dedicated digital integrated ecosystem

16:45 General discussion and conclusions, Prof. Santucci and Prof. Gallagher

A parallel AKU patient meeting will run after lunch in room 13 (ground floor).

Participation is free; please register at this link

<https://goo.gl/forms/M6EJSjYnOzEEqch2>