

11th International Workshop on Alkaptonuria (AKU)

University of Siena, Dept. Biotechnology, Chemistry and Pharmacy
via Aldo Moro 2 (room 14, 1st floor)
Wednesday 30th January 2019, 9:30 – 17:00

Participation is free; please register at this link
<https://goo.gl/forms/M6EJSjYnOzEEngch2>



9:00 Registration
9:30 Welcome message from
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) PDTA (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 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 Jebarai (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 (provisional title)

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)