



DIPARTIMENTO DI BIOTECNOLOGIE,
CHIMICA E FARMACIA
Department of Excellence 2018-2022



BiBiM 2.0 – Regional Executive
Pegaso PhD School in Biochemistry
and Molecular Biology



UNIVERSITÀ
DI SIENA
1240



ASSOCIAZIONE ITALIANA MALATI ALKAPTONURICI



Azienda ospedaliero-universitaria Senese



11th International Workshop on Alkaptonuria (AKU)

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

Provisional programme

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

- 9:30 Welcome message from Prof. **Francesco Frati**, *Rector*, and Prof. **Annalisa Santucci**, *Head of Dept. Biotechnology, Chemistry and Pharmacy – Dept. of Excellence 2018-2022*
- 9:40 Welcome message from Dr. **Valtere Giovannini** (*General Director of Siena University Hospital, Regional Center for Precision Medicine*), and Prof. **Antonio Federico** (*HCP Representative for MetabERN, Corporate and Regional Coordinator for Rare Diseases, Siena University Hospital*)
- 9:50 Dr. **Andrea Paolini** (*Toscana Life Sciences*) and Dr. **Andrea Frosini** (*UVaR, Regione Toscana*)
Regional policy on rare diseases & Precision Medicine

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 Dr. Albina Tummolo
(Bari Hospital) Experience with AKU patients at Bari Hospital (*provisional title*)

11:00 Coffee break

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

- 11:30 Dr. Wendy Inrone
(NIH, USA) Alkaptonuria in the United States
- 11:45 Hollie Washington
(RLBUH Liverpool, UK) UK National Alkaptonuria Centre and logistics
- 12:00 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





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| 12:15 | Dr. Mariusz Kujawa
(Medical University of Gdansk, Poland) | Care for paediatric patients with alkaptonuria |
| 12:30 | Prof. Maurizio Scarpa, Prof. Sylvia Sestini
(MetabERN) | MetabERN - European Reference Networks for hereditary metabolic disorders |

Light lunch

Session 3: AKU imaging

Chair: Prof. Gallagher

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| 14:00 | Dr. Leah Taylor
(RLBUH Liverpool, UK) | Assessment of AKU using quantitative image analysis |
| 14:15 | Dr. Alpesh Mistry
(RLBUH Liverpool, UK) | Interesting MRI findings in AKU and challenges in interpretation |
| 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

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| 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 and Dr. Lia Millucci
(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) | Prediction of residence time of novel HPPD inhibitors |
| 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

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| 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)

