

University Hospital Tuebingen,
Tuebingen, Germany

EuNet 
INNOCHRON

5th Management Committee Meeting 3rd Working Groups Meeting



Agenda

May 19-20, 2022,
Tuebingen, Germany



Universitätsklinikum
Tübingen

Day 1 - May 19

5th Management Committee Meeting [virtual/hybrid meeting only for MC Members]

08:45 - 09:00 (Action's Chair/Vice-Chair, GH Manager)

09:00 - 11:00 Management Committee Meeting

11:00 - 11:15 Coffee Break

3rd Working Groups Meeting – PART I [face-to face/hybrid meeting, all participants]

11:15 - 11:30 Welcome to all participants (J. Skokowa, H. Papadaki)

11:30 - 13:30 WG1 Session: Congenital Neutropenias (Chairs: A. Warren, K. Welte)

- ADAN: ACKR1/DARC associated neutropenia (J. Palmblad)
- Convergent somatic evolution from early life in a germline ribosomopathy (A. Warren)
- High-resolution structures of the human ribosome to decipher the molecular basis of snoRNAs dysfunction in disease (S. Pellegrino)
- WES/WGS of CN patients with unknown genetic disease cause (M. Klimiankou)

13:30 - 14:30 Lunch Break

14:30 - 16:30 WG2 Session: Acquired Neutropenias (Chairs: P. Höglund, J. Bux, F. Fioredda)

- Validation of a Luminex-based diagnostics tool for autoantibody diagnostics in childhood AIN (P. Höglund)
- Variants of unknown significance (VUS): use, throw away or keep in the “fridge”
 - VUS in NGS myeloid panel for CIN adult patients (G. Tsaknakis, H. Papadaki)
 - VUS found in long lasting and late onset neutropenia of children (F. Fioredda, Pietro Casartelli, M. Lanciotti)
 - VUS in CSF3R genic or VOUS. (O. Steinberg, H. Tamary)
- Update on guidelines diagnostic algorithms (F. Fioredda - H. Papadaki)
- Acquired neutropenia in children: epiphenomena of immunedysregulation (A. Beccaria)
- WES in CIN - a first approach (A. Grossi - G. Tsaknakis)

16:30 - 16:45 Coffee Break

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Day 1 - May 19

16:45 - 17:30 Key-Note Speaker: "The history of congenital neutropenia" (Karl Welte)

17:30 - 18:00 Young EuNet-INNOCHRON

- A brief update and next steps (M. Klimiankou - V. Bezzeri)

18:00 - 19:00 Representation from the Industry and SMEs

- Genotype-Phenotype correlations in WHIM syndrome: studies of novel CXCR4 mutations (K. Zmajkovicova, X4 Pharmaceuticals)
- European Health Data & Evidence Network (EHDEN) (P. Natsiavas)

Networking Dinner

Day 2 - May 20

3rd Working Groups Meeting – PART II (face-to face/hybrid meeting to be attended by all participants)

09:00 - 11:00 WG3 Session: Mechanisms of Leukemic Evolution (Chairs: C. Mecucci, I. Touw)

- The epi-genomic landscape of monosomy 7 in adult MDS/AML (A. Lema Fernandez)
- Children with inherited cytopenia often has leukemia predisposition (H. Tamary)
- Evaluation of the leukemogenic role of acquired CSF3R and RUNX1 mutations in HAX1 congenital neutropenia using iPSC-based model of granulocytic differentiation (B. Dannenmann)
- Clonal metrics and dynamics in CIN (H. Papadaki, G. Tsaknakis)

11:00 - 11:15 Coffee Break

11:15 - 13:15 WG4 Session: Investigation of Targets for Novel Therapies (J. Skokowa, J. Cichy)

- Updates on generation iPSCs-based models to study involvement of inhibitors of neutrophil proteases in neutropenia (J. Cichy)
- CRISPR/Cas gene therapy of CN (J. Skokowa, M. Nasri, M. Ritter)
- De novo design of new granulopoietic proteins to treat neutropenia (M. ELGamacy)
- Potential novel agents in SDS treatment (V. Bezzeri)

13:15 - 14:15 Lunch Break

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Day 2 - May 20

14:15 - 16:15 WG5 Session: CNP Registries and Biobanking (C. Zeidler, K. Stamatopoulos)

- An update from the Training School of Thessaloniki (K. Stamatopoulos - C. Zeidler)
- Adoption of the SCNIR forms as official forms of EuNet-INNOCHRON (C. Zeidler)
- Data registration systems (E. Minga)
- CIN database (H. Papadaki)
- Perspectives for neutropenia study in Albania (A. Ivanaj)
- Perspectives for neutropenia study in Montenegro (A. Celebic)

16:15- 16:30 Coffee Break

16:30 - 17:00 Patient Organizations: SDS Germany (D. Schorling, H. Blöcker)

17:00 - 17:30 An update on the Covid project (M. Spanoudakis, J. Palmbad)

17:30 - 17:45 An update from the Gender Equality Team (J. Roganovic - D. Yilmaz Karapinar)

17:45 - 18:30 All: Discussion for further steps

18:30 Closing remarks (J. Skokowa, H. Papadaki)

EuNet-INNOCHRON | CA18233

European Network for Innovative Diagnosis and Treatment of Chronic Neutropenias

COST | European Cooperation in Science and Technology



Local Organiser

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Speakers/Chairs	Affiliation	City	Country
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Valentino Bezzeri	Cystic Fibrosis Center, Azienda Ospedaliero Universitaria Ospedali Riuniti di Ancona	Ancona	Italy
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Karl Welte	University Children's Hospital Tuebingen	Tübingen	Germany
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Katarina Zmajkovicova	X4 Pharmaceuticals (Austria) GmbH	Vienna	Austria

*Speakers/Chairs in alphabetical Order



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