



This European Academy of Andrology (EAA) Training School in Genetics has been co-organized with the Andronet COST Action CA20119

Scope and purpose: EAA School in Genetics aims to offer the high-quality state-of-the-art level training to physicians and biomedical researchers interested in medical genetics with the specific focus on the genetics of andrological conditions. This will increase the knowhow and broader application of genetic testing among the andrologists, speed up the transfer of knowledge concerning recent advances in genetics and standardization of the utility and interpretation of novel genetic tools in the clinical practice. The course is also expected to promote genetic research in European andrology centres, with the specific focus on promoting synergetic and complementary actions.

Language: English

Number of EAA credits: 2

Participants: 21

EAA 2nd School in Genetics 2025, Tallinn, Estonia

Location, time: Conference Centre of Andrology Clinic in Tallinn, Tartu University Hospital Estonia; 22-24.01 2025

Organizers:

Maris Laan, PhD; professor of human genetics, Vice-Dean for research, Faculty of Medicine, University of Tartu

Kristjan Pomm, MD; Head of the Andrology Clinic, Tartu University Hospital

International faculty:

Aleksander Giwercman, MD, PhD; professor; Dept. of Translational Medicine, Lund University; the Chairman at Fertility Centre, Malmö University Hospital, Sweden

Frank Tüttelmann, MD; Director, Centre of Medical Genetics, Institute of Reproductive Genetics, University of Münster, Germany.

Stephane Viville, PhD; professor, Institut de génétique et de biologie moléculaire et cellulaire (IGBMC), Strasbourg, France

Kristian Almstrup, PhD; Senior Scientist, Dept. of Growth and Reproduction, Copenhagen University Hospital – Rigshospitalet; Associate Professor, University of Copenhagen, Denmark

Miguel Xavier, PhD; Senior Research Associate, Biosciences Institute, Newcastle University, UK

Fredrik Wiklund, PhD; Senior lecturer, Department of Medical Epidemiology and Biostatistics, Karolinska Institute, Stockholm, Sweden

Antonio Capalbo, PhD; Chief Scientific Officer, Juno Genetics; Assistant Professor, Unit of Medical Genetics, Center for Advanced Studies and Technology (CAST), "G. d'Annunzio" University of Chieti-Pescara, Italy

Course manager: Mailis Sütt (mailis.sutt@kliinikum.ee)

Course content and objective: The course aimed to revise the basic concepts of medical genetics, provide the state-of-the-art overview of currently applied genetic tools in the andrology clinic and introduce the current advances in androgenetics and their perspective clinical impact.

Programme and materials:

Topics covered:

- Human genome variation – chromosomal, monogenic and complex diseases
- Practical exercises 1 – monogenic disorders (in two parallel groups) Interactive discussion on clinical cases with a genetic cause, and their clinical assessment
- Practical exercises 2 – chromosomal disorders (in two parallel groups) Interactive discussion on clinical cases with a genetic cause, and their clinical assessment
- Genomic technologies in advancing molecular diagnostics
- From diagnostic Y-chromosome testing to advanced molecular diagnostics of monogenic male infertility – Münster experience
- Medically assisted reproduction – preconception carrier screening and preimplantation genetic testing
Sperm DNA defragmentation index – methods and clinical value
- Involvement of long and short non-coding RNAs in the pathogenesis of spermatogenic failure and modulating Klinefelter testicular phenotype
- Monogenic causes of female infertility and shared etiology with male infertility. A new paradigm in MAR practice
- Advances of genetics in infertility management – Estonian experience in translating basic research outcomes to the clinic
- Role of de novo mutations in male infertility – Newcastle experience
- Ethical dilemmas and challenges in andrology in the era of advanced genetic testing
- Opportunities, rationale and clinical benefit of advanced genetic testing in the efficient management of an infertile couple
- Genetics of testicular cancer
- Germline and somatic genetics of prostate cancer

As part of our program, all the participants were required to complete two online lectures before the course as homework:

Genetics in Medicine (55 minutes long)

<https://ut.cloud.panopto.eu/Panopto/Pages/Viewer.aspx?id=dfaa0b4f-4114-411e-a4ee-b2560072dbd0>

Genetic Variation (24 minutes long)

<https://ut.cloud.panopto.eu/Panopto/Pages/Viewer.aspx?id=9d18d671-d7d8-4b9b-85c2-b25600728413>

These lectures were designed to provide a brief overview of the fundamental concepts in the field of genetics. After completing these lectures, it was required to take a complementary test to help assess the knowledge of the basic genetics terminology.

<https://sisu.ut.ee/eaa/genetics-terminology-tests/>

Participants: The attendees of the second course represented Sweden, Turkey, Portugal, UK, Cyprus, Italy, Romania, Denmark, Indonesia, Germany, Estonia, Cyprus

Participants have variable background: clinical andrologist, physicians in other reproductive medicine area (e.g. ART), MD, PhD or PhD students (andrology, genetics), molecular geneticist, andrology lab technicians, and cytogeneticist.

Feedback: The participants liked the structure of the course. All the lectures and speakers received positive feedback and were identified as highly relevant and educating to the attendees. Discussions on clinical cases with a suspected or diagnosed genetic cause brought along by the participants received a very good feedback and were expressed to be highly insightful. Also the city tour and dinner at a medieval restaurant added spark to the course. The overall feedback was very positive and supported regular organization of this EAA School.

- Overall an excellent course in terms of organisation, content, location. You could feel that you were welcome. Excellent discussions and an atmosphere was there that allowed questions and discussions.
The course was exceptionally organized, starting with the pre-course arrangements and thorough instructions
- Prof. Maris Laan is a strong expertise in her area of specialization. Genetics is a complicated subject, yet having an excellent teacher can help you grasp it more easily. Maris Laan managed not only to instruct us in a very practical way, but also to motivate us to delve deeper into genetics in the future.
- Very good speakers. Every topic was interesting.
- The course was very well organised, interesting talks, very interesting practical exercises (cases)

COURSE PROGRAMME

Wednesday, January 22th

13:00 Maris Laan, Opening remarks

13:15 – 14:30 **Maris Laan** “Human genome variation – chromosomal, monogenic and complex diseases”

14:30 – 14:45 Coffee break

14:45 – 16:00 Practical exercises 1 – monogenic disorders (in two parallel groups; **Rain Inno, Kristiina Lillepea, Anna-Grete Juchnewitch & Maris Laan**)

16:00 – 16:15 Coffee break

16:15 – 17:00 **Miguel Xavier** “Genomic technologies in advancing molecular diagnostics”

17:00 – 18:15 Practical exercises 2 – chromosomal disorders (in two parallel groups; **Rain Inno, Kristiina Lillepea, Anna-Grete Juchnewitch & Maris Laan**)

19:00 Welcome reception – dinner (Hotel Mercure)

Thursday, January 23rd

9:00 – 10:30 **Frank Tüttelmann** “From diagnostic Y-chromosome testing to advanced molecular diagnostics of monogenic male infertility – Münster experience”

10:30– 10:45 *Coffee break*

10:45 – 12:15 **Antonio Capalbo** “Medically assisted reproduction – preconception carrier screening and preimplantation genetic testing” (*Virtual lecture*)

12:15 – 13:15 *lunch*

13:15 – 14:15 **Aleksander Giwercman** “Sperm DNA defragmentation index – methods and clinical value”

14:15 – 15:15 **Kristian Almstrup** “Involvement of long and short non-coding RNAs in the pathogenesis of spermatogenic failure and modulating Klinefelter testicular phenotype.”

15:15 – 18:00 guided tour Tallinn Old City

18:30 – ... Dinner in Old City (restaurant **Olde Hansa**, <https://www.oldehansa.ee/our-menus/>)

Friday, January 24th

9:00 – 10:30 **Stephane Viville** “Monogenic causes of female infertility and shared etiology with male infertility. A new paradigm in MAR practice.”

10:30-10:45 Coffee break

10:45 – 12:00 **Maris Laan** “Advances of genetics in infertility management – Estonian experience in translating basic research outcomes to the clinic”

12:00 – 12:45 lunch

12:45-13.10 – Visiting upstairs Andrology Clinic and Genetics and Personal Medicine Center

13:10 – 13:45 **Miguel Xavier** “Role of de novo mutations in male infertility – Newcastle experience”

13:45 – 14:45 Discussions led by **Margus Punab & Kristjan Pomm**

“Ethical dilemmas and challenges in andrology in the era of advanced genetic testing”

“Opportunities, rationale and clinical benefit of advanced genetic testing in the efficient management of an infertile couple”

15:00– 15:30 Coffee break

15:30 – 16:00 **Kristian Almstrup** “Genetics of testicular cancer”

16:00 – 17:00 **Fredrik Wiklund** “Germline and somatic genetics of prostate cancer”

17:00 – 17:30 *Wrap-up* discussions on the actions needed to promote advanced genetic testing in European andrology centres, the challenges in test standardization, data interpretation and counselling of patients.

Farewell!





The course was supported by the European Academy of Andrology, Andronet COST Action CA20119 and the Andrology Clinic of Tartu University Hospital. COST Andronet supported the participation of 3 participants and EAA provided 5 travel grants.

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