European Academy of Andrology (EAA) School in Genetics

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: ~30

EAA 1st School in Genetics 2019, Tartu, Estonia

Location, time: Conference Centre of University of Tartu Library, Tartu, Estonia; 11-13.09 2019
Organizers: Prof. Maris Laan (Chair of human genetics, Institute of Biomedicine and Translational Medicine, University of Tartu) & Prof. Margus Punab (Andrology Centre, Tartu University Hospital); EAA Tartu Centre

Faculty: Prof Maris Laan, Prof Margus Punab, Dr. Pille Hallast, Dr. Lili Milani (Tartu, EST), Prof Frank Tüttelmann (Münster, GER), Prof Joris Veltman (Newcastle, UK), Prof Csilla Krausz (Florence, IT)
Practical course: Prof. Maris Laan, Drs. Laura Kasak, Rain Inno, Triin Kikas (Tartu, EST)
The course manager: Ms 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*:** A blend of lectures, seminars based on home work and practical course on using human genome databases (See: *Programme*). Course materials included seminar/lecture slides shared with the course participants and faculty after the course scientific articles on medical genetics and androgenetics, molecular diagnostic methods and human genome databases accompanying the lectures.

Topics covered:

- Human genome and its variation, genomic technologies; terminology in medical genetics
- From cytogenetics defects to Mendelian inheritance of genetic disease
- Classical and molecular cytogenetics in andrology workup; focus on Klinefelter syndrome
- Diagnostic Y-chromosome testing and external quality control of the EMQN/EAA
- Y-chromosomal analysis beyond AZF regions – relevant or not in andrology?
- Clinical considerations and applications of WES and WGS in andrology research and clinic
- De novo mutations – frequent or rare cause of male infertility?
- Overlap between the genetics of infertility and cancer
- Digenic/oligogenic effects – role in andrological conditions?
- The resources of biobanks for the research in clinical genetics

- Introduction to pharmacogenetics and its potential in andrology clinic
- Human medical genetics databases and their utility in clinical practice
- Interactive discussion on clinical cases with a genetic cause, and their clinical assessment

Participants: The attendees of the first course represented Denmark, Belgium, Spain, Croatia, Latvia, Moldavia, Estonia. Participants have variable background: clinical andrologist, physicians in other reproductive medicine area (e.g. ART), MD, PhD or PhD students (in pediatrics, andrology, genetics), molecular geneticists and cytogeneticist.

Feedback: The participant liked the overall structure of the course, mixing lectures and practical sessions. All the lectures and speakers received positive feedback and were identified as highly relevant and educating to the attendees. The organizers' 'experiment' to initiate interactive presentations and discussions on clinical cases with a suspected or diagnosed genetic cause brought along by the participants received a very good feedback and was expressed to be highly insightful. The overall feedback was very positive and supported regular organization of this EAA School.

PROGRAMME „EAA School in Genetics 2019“

Wednesday, September 11th

Lectures ‘Basic principles in medical genetics’

9.00 – 9.10 **Maris Laan** ‘Opening remarks – era of genomic medicine in andrology?’
9.10 – 10.10 **Joris Veltman** “Human genome, genetic variation, genomic technologies”
10.10 – 11.10 **Frank Tüttelmann** “From cytogenetics defects to Mendelian inheritance of genetic disease”

11.10 – 11.40 coffee break

11.40 – 13.00 Interactive seminar in 2 parallel groups “Revisiting the basic terminology and principles in medical genetics” Faculty: **Maris Laan & Laura Kasak, Rain Inno & Triin Kikas**

13.00 – 13.45 Lunch

13.45 – 15.30 Guided walk to the Tartu Dome Hill and University of Tartu Museum (guide: **Anu Rae**)

15.30 – 18.00 **Practical courses:**

- Group 1: Hands-on learning of human medical genetics databases and their utility in clinical practice (computer class)

Faculty: Rain Inno, Laura Kasak & Triin Kikas

- Group 2: Interactive presentation and discussion on clinical cases with a suspected or diagnosed genetic cause, and their clinical assessment

Faculty: Margus Punab, Maris Laan, Csilla Krausz, Frank Tüttelmann, Joris Veltman

Dinner for the course lecturers*: restaurant ‘Ülikooli Kohvik’ (Ülikooli 20) at 19:00*

Thursday, September 12th

Lectures ‘Androgenetics -state-of-the-art and respective clinical utility’

9.00 – 9.30 **Margus Punab** “Clinicist's view to the status of genetic testing in andrology clinic”
9.30 – 10.00 **Csilla Krausz** “Diagnostic Y-chromosome testing and external quality control of the EMQN/EAA”
10.00 – 10.45 **Joris Veltman** “Clinical considerations and current applications of using whole exome and genome sequencing in andrology research and clinic”

10.45 – 11.15 coffee break

11.15 – 11.45 **Frank Tüttelmann** “Utility and diagnostic yield of classical and molecular cytogenetics for the detection on gross and submicroscopic chromosomal aberrations in andrology workup”

11.45 – 12.15 **Csilla Krausz** "Klinefelter syndrome from a broad prospective"
12.15 – 13.00 **Maris Laan** "Digenic/oligogenic effects – role in andrological conditions?"

13.00 – 14.00 Lunch

14.00 – 16.30 **Practical courses:**

- Group 2: Hands-on learning of human medical genetics databases and their utility in clinical practice (computer class)

Faculty: Rain Inno, Laura Kasak & Triin Kikas

- Group 1: Interactive presentation and discussion on clinical cases with a suspected or diagnosed genetic cause, and their clinical assessment

Faculty: Margus Punab, Maris Laan, Csilla Krausz, Frank Tüttelmann, Joris Veltman

Bus leaving to Mooste at 17:00 (travel 45 min)

17.45 – 22.00 Course Dinner and Tour in Mooste Manor (http://www.moostemois.ee/?keel=eng)

Friday, September 13th

Lectures 'Current research advances in androgenetics'

9.00 – 9.45 **Frank Tüttlemann** "Interdisciplinary approach towards male infertiliy - project 'Male Germ Cells: from Genes to Function'
9.45 – 10.15 **Joris Veltman** " De novo mutations – frequent or rare cause of male infertility?"
10.15 – 11.00 **Pille Hallast** "Y-chromosomal analysis beyond AZF regions – relevant or not in andrology?"

11.00 – 11.30 coffee break

11.30 – 12.00 **Csilla Krausz** "Overlap between the genetics of infertility and cancer"
12.00 – 12.45 **Lili Milani** (Estonian Biobank, visiting Lecture) "The resources of biobanks for the research and pharmacogenic applications in clinical genetics".

12.45 – 13.30 Lunch

13.30 – 15.00 **Wrap-up session:**

- Course faculty (led by **Csilla Krausz**) 'Feedback from the interactive case discussions'
- Interactive discussion (all) on the actions needed to promote and standardize advanced genetic testing and interpretation/counselling of its outcomes in European andrology centres
- **Margus Punab** "Closing remarks"