Semmelweis University

Institute of Genomic Medicine and Rare Disorders

The contemporary role of genetic testing in rare disorders

The preceptorship program of the
Institute of Genomic Medicine and Rare Disorders
01-02 February 2024
Budapest, Hungary

The course will cover the different genetic tests and technologies available for diagnosing rare diseases, such as PCR, real-time PCR, MLPA-based technologies, whole exome sequencing, and whole genome sequencing. We will discuss the clinical applications of genomics. This topic will focus on the use of genomics in clinical practice, including the interpretation of genetic test results, genetic counseling, and the integration of genomics into patient care. We will discuss the importance of rare disease registries and databases for improving the diagnosis and treatment of rare diseases, as well as the challenges and opportunities associated with data sharing and privacy. In the seminar about precision medicine and targeted therapies, we will explore the potential of precision medicine and targeted therapies for treating rare diseases, including the use of gene therapy, cell therapy, and small-molecule drugs. Finally, we will address the ethical, legal, and social implications of clinical genetics and genomics in the diagnosis of rare diseases, including issues related to informed consent, genetic discrimination, and access to genetic testing and treatment.

We would like to kindly offer the participants if they have interesting cases they can share with the program fellows during the course. So, if there is a case where genetic testing or its interpretation posed a challenge, there will be an opportunity to present and discuss it on three slides. Our only request is that you kindly send the three slides for presentation a week before the program so that we can incorporate them into the program.

Program

1. Day: 01 February 2024
Location: Semmelweis Salon 1085 Budapest Üllői Str 26. 1. Floor

8.30 – 8.40
Introduction - Maria Judit Molnar

8.40 -10.10
The role of clinical genetics/genomics in the rare disease diagnostic (introduction to the topic) - Maria Judit Molnar

10.10- 10.30
Coffee break

10.30 – 12.00
Databases, ontologies, and the incorporation of web-based solutions in developing of medical hypotheses and clinical decision making (OMIM, ClinVar, DECIPHER, Orphanet, HPO, phenotype-driven gene prioritization) Workshop – Viktor Molnar

12.00 - 12.30 The potentials and pitfalls of precision medicine in 2024 – György Nemeth

12.30 - 13.30 Lunch

Lab Visits
Location: Semmelweis University Rokus Block 1085 Budapest Gyulai Pal Str 2.

13.30 - 15.00
Genetic tests frequently used in the rare disease diagnostic I. (1.) PCR, PCR+RFLP Sanger Sequencing, 2.) real time PCR, MLPA, 3.) Next generation sequencing - Aniko Gal, Szabolcs Udvari, Klara Pentelenyi

15.00 – 16.30
Challenges of the genetic testing (workshop in the lab including your case presentation as well) - Peter Balicza, Viktoria Szabo, Szabolcs Udvari

16.30 – 16.50
Coffee break

16.50 – 18.20
How to unlock genomic data? (workshop in small groups: variant calling, clinical interpretation, VUS, secondary findings – including your cases as well) - Viktor Molnar, Peter Balicza, Tamas Szlepak, Viktoria Szabo

2. Day: 02 February 2024
Location: Semmelweis Salon 1085 Budapest Üllői Str 26. 1. Floor

8.30 - 10.00
Personalized therapeutic decisions in clinical practice - Team competition in small groups.
Facilitators: Viktor Molnar, Viktoria Szabo, Noemi Varga

10.00- 10.15
Biobreak

10.15- 11.45
Genetic counselling in the era of broad genetic testing (workshop including your cases as well) - Marta Szegedi, Barbara Csendes

11.45-12.05
The importance of NBS screening – Ildiko Szatmari

12.05 – 12.25
How to build biobanks and rare disease registries – Idris Jimoh

12.20.- 12.45
The role of the real-world data collection and data sharing in rare disorders – Peter Balicza

12.55.- 13.20
How to improve your diagnostic skills – Maria Judit Molnar

13.20. Closing remarks

13.20- 14.00
Lunch