

Genetics for Pediatric Neurologists Faculty			
Co-chairs: Andrea Nemeth, Dana Craiu			
Lecturers - Dana Craiu, Andrea Nemeth, Andreas Brunklaus, Enrico Bertini, Nadia Bahi-Buisson and Anna Jansen			
Day 1: Room C5			
08.00 – 08.30	30 min	Registration	
08.30 – 09.00	30 min	Housekeeping, introduction. Pre-course questions	Dana Craiu/ Andrea Nemeth
09.00 – 09.30	30 min	Basic genetics 1 Chromosomes and DNA, RNA and Protein Concept of resolution Basic and more complex inheritance patterns (Mosaicism somatic and germline), reduced penetrance	Andrea Nemeth/ Dana Craiu
09.30 – 10.15	45 min	Basic genetics 2 Types of Genetic testing: karyotypes (and when useful in 2026), arrays, PCR, sequencing (NGS - panels, exome, genome) Complex testing: methylation, X inactivation, radiation sensitivity etc	Andrea Nemeth/Dana Craiu
10.15 – 10.45	30 min	Break	
10.45 – 11.30	45 min	From data to interpretation The importance of accurate phenotyping with examples, Reference sequences, MAF, Filtering, pathogenicity, VUS's, resources, ACMG guidelines	Dana Craiu/ Andreas Brunklaus
11.30 - 12.15	45 min	Principles of Gene Therapy; successes and failures	Enrico Bertini
12:15 - 13:00	45 min	Ethical issues Genetic testing and consent - testing minors, incidental findings, non-paternity, challenges of implementing newborn screening	Anna Jansen
13.00 – 14.00		Lunch	
14.00 - 14.45	45 min	Diagnostic yields, missing diagnoses Next steps with negative cases -Interactive session with participants 3-4 cases	Faculty
14:45 - 15.30	45 min	Examples of difficult to interpret cases	Dana Craiu
15.30 – 16.00		Break	
16.00 – 16.45	45 min	Future developments - Diagnostics Long reads, RNA Rt-PCR RNA seq Episignatures, Optical Genome Mapping (OGM), multiomics	Faculty
16.45-18.00	1 hour 15 min	Case presentations by the participants Room C5 / Room C3 / Room C1 /Room 6	
18.00		Conclusions	

Day 2: Room C5			
09.00 - 09.45	45 min	Neuro-muscular	Enrico Bertini
09.45 - 10.30	45 min	Genetic Causes of Neurodevelopmental Disorders in Clinical Practice	Anna Jansen
10.30 - 11.00		Break	
11.00 - 12.00	1 hour	Case-presentations by the participants Room C5 / Room C7A / Room C7B	
12.00 - 12.45	45 min	Epilepsies	Andreas Brunklaus
12.45 - 13.30	45 min	Movement disorders	Nadia Bahi- Buisson
13.30 - 14.45	75 min	Lunch	
14:45 - 15.30	45 min	Malformations	Anna Jansen
15.30 - 16.15	45 min	Mitochondrial Disease	Enrico Bertini
16.15 - 17.00	45 min	Break (+ faculty discussions of the best presentation)	
17.00 - 17.15	30 min	Best presentation award	Dana Craiu
17.15 - 17.30		Conclusions	All