

**EPNS Training Course: Ferrara, Italy, 25-26 May 2026**  
**Genetics for Paediatric Neurologists - Preliminary programme**



<b>Genetics for Pediatric Neurologists Faculty</b>			
Co-chairs: Andrea Nemeth, Dana Craiu			
Lecturers - Dana Craiu, Andrea Nemeth, Andreas Brunklaus, Enrico Bertini, Nadia Bahi-Buisson and Anna Jansen			
Day 1			
<b>08.00 – 08.30</b>	<b>30 min</b>	<b>Registration</b>	
<b>08.30 – 09.00</b>	<b>30 min</b>	<b>Housekeeping, introduction. Pre-course questions</b>	<b>Dana Craiu/ Andrea Nemeth</b>
<b>09.00 – 09.30</b>	<b>30 min</b>	<b>Basic genetics 1</b> Chromosomes and DNA, RNA and Protein Concept of resolution Basic and more complex inheritance patterns (Mosaicism somatic and germline), reduced penetrance	<b>Andrea Nemeth/ Dana Craiu</b>
<b>09.30 – 10.15</b>	<b>45 min</b>	<b>Basic genetics 2</b> 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	<b>Andrea Nemeth/Dana Craiu</b>
<b>10.15 – 10.45</b>	<b>30 min</b>	<b>CB</b>	
<b>10.45 – 11.30</b>	<b>45 min</b>	<b>From data to interpretation</b> The importance of accurate phenotyping with examples, Reference sequences, MAF, Filtering, pathogenicity, VUS's, resources, ACMG guidelines	<b>Dana Craiu/ Andreas Brunklaus</b>
<b>11.30 - 12.15</b>	<b>45 min</b>	<b>Principles of Gene Therapy; successes and failures</b>	<b>Enrico Bertini</b>
<b>12:15 - 13:00</b>	<b>45 min</b>	<b>Ethical issues</b> Genetic testing and consent - testing minors, incidental findings, non-paternity, challenges of implementing newborn screening	<b>Anna Jansen</b>
<b>13.00 – 14.00</b>		<b>Lunch</b>	
<b>14.00 - 14.45</b>	<b>45 min</b>	<b>Diagnostic yields, missing diagnoses</b> <b>Next steps with negative cases -</b> Interactive session with participants <b>3-4 cases</b>	<b>Faculty</b>
<b>14:45 - 15.30</b>	<b>45 min</b>	<b>Examples of difficult to interpret cases</b>	<b>Dana Craiu</b>
<b>15.30 – 16.00</b>		<b>CB</b>	
<b>16.00 – 16.45</b>	<b>45 min</b>	<b>Future developments - Diagnostics</b> Long reads, RNA Rt-PCR RNA seq Episignatures, OGM - Bionano, Multiomics	<b>Faculty</b>
<b>16.45-17.45</b>	<b>1 hour</b>	<b>Case-presentations by the participants</b>	
<b>19.30 - 22.30</b>		<b>Social event, including dinner</b>	

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<b>Day 2</b>			
<b>09.00 - 09.45</b>	<b>45 min</b>	<b>Neuro-muscular</b>	<b>Enrico Bertini</b>
<b>09.45 - 10.30</b>	<b>45 min</b>	<b>Genetic Causes of Neurodevelopmental Disorders in Clinical Practice</b>	<b>Anna Jansen</b>
<b>10.30 - 11.00</b>		<b>CB</b>	
<b>11.00 - 11.45</b>	<b>45 min</b>	<b>Mitochondrial Disease</b>	<b>Enrico Bertini</b>
<b>11.45 - 12.30</b>	<b>45 min</b>	<b>Epilepsies</b>	<b>Andreas Brunklaus</b>
<b>12.30 - 13.15</b>	<b>45 min</b>	<b>Movement disorders</b>	<b>Nadia Bahi- Buisson</b>
<b>13.15 - 14.30</b>	<b>75 min</b>	<b>Lunch</b>	
<b>14:30 - 15.15</b>	<b>45 min</b>	<b>Malformations</b>	<b>Anna Jansen</b>
<b>15.15 - 16.15</b>	<b>1 hour</b>	<b>Case-presentations by the participants</b>	
<b>16.15 - 17.00</b>	<b>45 min</b>	<b>CB (+ faculty discussions of the best presentation)</b>	
<b>17.00 - 17.15</b>	<b>30 min</b>	<b>Best presentation award</b>	<b>Dana Craiu</b>
<b>17.15 - 17.30</b>		<b>Conclusions</b>	<b>All</b>