



**Università  
degli Studi  
di Ferrara**

The European Reference Network for Rare Neuromuscular Diseases Euro-NMD welcomes you on November 6<sup>th</sup>, 7<sup>th</sup> & 8<sup>th</sup>, 2019 to the 3<sup>rd</sup> Annual Meeting held at the University Hospital Cona in Ferrara, Italy  
Azienda Ospedaliero Universitaria di Ferrara

WEDNESDAY, NOVEMBRE 6 <sup>TH</sup>					
Aula Magna					
SATELLITE MEETING with the ESHG on GENETIC TESTING					
<i>Chairpersons: Alessandra Ferlini, Serenella Servidei</i>					
14.30-15.00	Introduction, objectives of the WS <i>Prof. Alessandra Ferlini (Ferrara, Italy)</i>				
15.00-15.30	Gene variant annotation approaches <i>Prof. Maurizio Genuardi (Rome, Italy)</i>				
15.30-16.00	WGS Diagnostic Translation <i>Hans Scheffer (Nijmegen, The Netherlands)</i>				
16.00-16.30	Coffee break				
16.30-17.00	« FSHD from epigenetic mechanisms to therapy » <i>Dr. Julie Dumonceaux (London, UK)</i>				
17.00-17.30	Discussion et Conclusions, Photo Call <i>Prof. Alessandra Ferlini (Ferrara, Italy)</i>				
PARALLEL MEETINGS					
	Aula Magna	Room 1	Room 2	Room 7	Room 8
17.30-19.00				Mitochondrial Diseases <i>Rita Horvath</i>	
20.00	Speaker's Dinner ( <i>Invitation only</i> )				

THURSDAY, NOVEMBER 7 <sup>TH</sup>					
PARALLEL MEETINGS					
	Aula Magna	Room 2	Room 4	Room 7	Room 8
09.00-10.30	CPMS <i>Chloe Blewitt</i>	Imaging <i>Pierre Carlier</i>	Muscle <i>Jens Schmidt</i>	Neurophysiology <i>Camiel Verhamme</i>	Education <i>Andoni Urtizberea</i>
10.30-11.00	Coffee break				

THURSDAY, NOVEMBER 7 <sup>TH</sup>	
ERN ANNUAL MEETING Day 1	
11.00-11.15	Welcome <i>Alessandra Ferlini/Teresinha Evangelista/Professor Patrizio Bianchi</i>
SESSION 1	<i>Chairpersons : Alessandra Ferlini, Luisa Politano</i>
11.15-11.45	Ramblings in myology: My first 60 years <i>Victor Dubowitz (London; UK)</i>
11.45-12.05	Newborn Screening (Patients point of view) <i>François Lamy (Paris, France)</i>
12.05-12.25	Pilot Newborn screening example (SMA) <i>Eduardo Tizzano (Barcelona, Spain)</i>
12.25-12.45	German experience on screening more than 250.000 new-borns for SMA <i>Astrid Blaschek (Munich, Germany)</i>
12.45-13.45	LUNCH
SESSION 2	<i>Chairpersons : Bjarne Udd, Anna Sarkozy</i>
13.45-14.10	WGS in the genetics diagnostic lab <i>Hans Scheffer</i>
14.10-14.30	eNMD aims & meeting report <i>Gabriele Siciliano (Pisa, Italy)</i>
14.30-15.30	Case reports – 10 min. each + 5 min. discussion <i>Dr. Guido Primiano, Pr. Andrés Nascimento, Pr. Jens Schmidt, Dr. Ivana Babic Bozovic</i>
15.30-16.00	Coffee break, photo call
16.00-18.00	BOARD MEETING <span style="float: right;">Aula Magna</span>
18.00-19.30	ERN EXECUTIVE COMMITTEE MEETING <span style="float: right;">Aula Magna</span>
20.30	Social Dinner in Ferrara (Estense Duke Castle)

FRIDAY, NOVEMBER 8<sup>TH</sup>

## ERN ANNUAL MEETING Day 2

**SESSION 3**     *Chairpersons: Davide Pareyson, Teresinha Evangelista*

**08.30-09.15**     *Solve-RD & what we can learn from other ERN's  
Holm Grässner (Tübingen, Germany)*

**09.15-09.45**     *NMD-related Projects (EJP-RD) Dr. Eleonora Passeri*

**09.45-11.00**     *Case reports - 10 min. each + 5 min. discussion Dr. Davide Ognibene, Dr. Fernanda Fortunato, Dr. Michaela Catteruccia, Dr. Giulia Ricci, Dr. Viktor Molnar*

**Photo Call, end of the meeting**

**11.00-11.30**     **Coffee break**

### PARALLEL MEETINGS

	Aula Magna	Room 1	Room 2	Room 7	Room 8
<b>11.30-13.00</b>	<b>Pathology</b> <i>Teresinha Evangelista</i>		<b>Peripheral Nerve</b> <i>D. Pareyson</i>	<b>Genetics</b> <i>Alessandra Ferlini</i>	
<b>13.00- 14.30</b>		<b>PAB</b> <i>F.Lamy</i>			



Estense Castle of Ferrara