

*** programme ***

Friday 13 December

14.00 - 15.00	Introductory session
14.00 - 14.10	Introduction and welcome, Alexandra Breukel, Managing Director ENMC
14.10 - 14.25	Welcome and introduction of participants, Anders Oldfors – Sweden\
14.25 - 14.40	Overview of muscle glycogen storage disorders, Pascal Laforêt - France
14.40 - 14.55	Overview of diseases with polyglucosan accumulation, Edoardo Malfatti - France
14.55 - 15.00	Discussion
15.00 - 15.30	Basic aspects on glycogen metabolism, Chair: Or Kakhlon
15.00 - 15.25	Glycogen metabolism in muscle and liver, Monique Piraud - France
15.25 - 15.30	Discussion
15.30 - 15.50	Coffee break
15.50 - 17.15	Basic aspects on glycogen metabolism, Chair: Or Kakhlon
15.50 - 16.20	Glycogenin as a primer for glycogen synthesis, Thomas Hurley - US
16.20 - 17.00	Glycogenin knock-out mouse model – biochemical aspects, Joan Guinovart - Spain
17.00 - 17.15	Discussion
17.15 - 17.45	Polyglucosan storage diseases, Chair: Or Kakhlon
17.15 - 17.45	Lafora disease: clinical aspects and molecular pathogenesis, Berge Minassian -Canada
17.45 - 17.55	Discussion
19.00	Dinner

Saturday 14 December

09.00 - 10.30	Polyglucosan storage diseases continued, Chair: Berge Minassian
09.00 - 09.15	Lafora disease: molecular pathogenesis, Jordi Duran - Spain
09.15 - 09.40	Branching enzyme deficiency (GBE1), Berge Minassian, Or Kakhlon
09.40 - 10.00	RBCK1-associated disease –clinical aspects, Elaine Murphy - UK
10.00 - 10.10	RBCK1-associated disease –pathological aspects, Anders Oldfors - Sweden
10.10 - 10.20	PRKAG2 cardiomyopathy, Karim Wahbi - France
10.20 - 10.30	Discussion
10.30 - 10.50	Coffee break
10.50 - 12.30	Glycogenin deficiency, Chair: Pascal Laforêt
10.50 - 11.00	Glycogenin-1 associated myopathy- clinical findings, Pascal Laforêt - France
11.00 - 11.10	Glycogenin-1 associated myopathy-pathological findings, Edoardo Malfatti - France
11.10 - 11.20	Glycogenin-1 associated cardiomyopathy - pathological findings, Anders Oldfors - Sweden
11.20 - 11.30	Genotype-phenotype correlation in glycogenin-1 deficiency, Carola Oldfors - Sweden
11.30 - 11.40	Glycogenin-2 in glycogenin-1 deficiency, Thomas Krag - Denmark/ Anders Oldfors - Sweden
11.40 - 12.00	Glycogenin-1 deficiency, is muscle glycogenolysis also affected? metabolism during exercise, John Vissing - Denmark
12.00 - 12.20	Discussion
12.20 - 13.00	Animal models of polyglucosan storage, Chair: Thomas Hurley
12.20 - 12.35	Equine polyglucosan storage disease, Richard Piercy - UK
12.35 - 12.50	Study of infrared microspectral signature of skeletal muscle from horses affected by polysaccharide storage myopathy, Marie-Anne Colle - France
12.50 - 13.00	Discussion
13.00 - 14.30	Lunch

EUROPEAN NEURO MUSCULAR CENTRE

14.30 - 15.30	Polyglucosan - structural and biochemical aspects, Chair: Matt Gentry
14:30 - 14.45	Histopathology aspects of polyglucosan myopathies, Edoardo Malfatti - France
14.45 - 15.00	Proteomic analysis of polyglucosan, Christer Thomsen - Sweden
15.00 - 15.20	Imaging-based high content screening for drug discovery, Miguel Weil - Israel
15.20 - 15.30	Discussion
15.30 - 16.00	Coffee break
16.00 - 18.00	Therapeutic aspects, Chair: John Vissing
16.00 - 16.25	Therapeutic approaches of APBD, Or Kakhlon - Israel
16.25 - 16.50	Polyglucosan bodies – enzymatic digestion, Matt Gentry - US
16.50 - 17.15	Using 'genetic modifiers' to discover safe and effective targets for the treatment of glycogenosis, Vyas Ramanan, Maze Therapeutics - US
17.15 - 17.40	Gene therapy approaches in GSDs and PB myopathies Federico Mingozzi - France
17.40 - 18.00	Discussion
19.00	Dinner

Sunday 15 December

09.00-09.20	Therapeutic perspectives, Chair: John Vissing
09.00-09.20	The patients' perspective, Camilla B. Lilleør - Denmark
09.20-11.00	Discussions and deliverables, Chair: Pascal Laforêt
09.20-09.50	Pathobiology future studies, Leader of discussion: Joan Guinovart
09.50-10.20	New diseases, collection of data and analyses Leader of discussion: Anders Oldfors
10.20-11.00	Treatment studies, Registries, outcome measures etc. Leader of discussion: John Vissing
11.00-11.30	Coffee break
11.30-12.00	Draft of the lay summary
12:00-13:00	Lunch and departure