

Programma ESN voorjaarssymposium

Dinsdag 7 april 2015

Vanaf 9.00	ONTVANGST EN REGISTRATIE
10.00-10.10	Welkom en Introductie Ans van der Ploeg, George Ruijter (Erasmus MC)
10.10-11.55	Innovatieve therapieën Sessievoorzitters: Ans van der Ploeg, George Ruijter
10.10-10.40	Gene Therapy: The End of the Beginning Rob Hoeben (LUMC, Leiden)
10.40-11.05	Predictive and therapeutic value of muscle stem cells for Pompe disease Pim Pijnappel (Erasmus MC, Rotterdam)
11.05-11.30	Gene- and Cell therapy for mitochondrial disorders René de Coo (Erasmus MC, Rotterdam / Maastricht UMC+)
11.30-11.55	Mitochondrial disease drug development from bench to bedside Jan Smeitink (Radboud UMC, Nijmegen)
11.55-12.45	LUNCH
12.45-14.15	VERGADERINGEN Biochemici EMZ, NVK sectie MZ, INVEST en MODAZ
14.15-15.30	Vrije voordrachten Sessievoorzitters: Jan Smeitink, Pim Pijnappel
	Stem cell based strategies for patients with liver disease Sabine Fuchs (UMC Utrecht)
	Lentiviral stem cell gene therapy for Pompe disease Qiushi Liang (Erasmus MC, Rotterdam)
	Identification of new metabolism disorders genes by integrative genomics methods Radek Szklarczyk (Maastricht UMC)
	Identification and characterization of aberrant splicing in Pompe disease using a generic approach. Atze Bergsma (Erasmus MC, Rotterdam)

Concensus recommendations on initiation and cessation of enzyme replacement therapy in patients with Fabry disease

Marieke Biegstraaten (AMC, Amsterdam)

- 15.30-16.00 **KOFFIE/THEE**
- 16.00-17.00 Huishoudelijke vergadering ESN
17.30 Sociale activiteit
- 20.00 **DINER**

Woensdag 8 april

- 9.00 -10.30 **Organoaciduriën en vetzuuroxidatiedefecten**
Sessievoorzitters: Monique Williams, Geert Martens
- 9.00-9.30 **Movement disorders and organic acidurias**
Tom de Koning (UMCG, Groningen)
- 9.30-10.00 **Organic acidemias - a personal up to date**
Stephanie Grünewald (University College, London)
- 10.00-10.30 **KOFFIE**
- 10.30-10.55 **New insights in the valine degradation pathway**
Sacha Ferdinandusse (AMC, Amsterdam)
- 10.55-11.40 **Vrije voordrachten**
Sessievoorzitters: Terry Derks, Jörgen Bierau
- Clinical severity in VLCAD deficient patients correlates with fatty acid oxidation flux in cultured fibroblasts: potential use for patients identified through NBS**
Jeanette Bleeker (AMC, Amsterdam / UMC Utrecht)
- First results from the EIMD registry on the phenotypic spectrum of organic acidurias (OA) and urea cycle defects (UCD) - on behalf of the European consortium**
Monique Williams (Erasmus MC)
- Markers for the prediction of disease severity in patients with mucopolysaccharidosis type IIIB**
Olga Meijer (AMC, Amsterdam)
- 11.40-12.40 **Uitreiking en presentaties Wadman-Van Gennip prijs en prijs beste artikel**

- 12.40-13.45 **LUNCH**
- 13.45-15.00 **Lysosomale stapelingsziekten**
Sessievoorzitters: Carla Hollak, Ans van der Ploeg
- 13.45-14.05 **Cell Therapy in Lysosomal Storage Diseases: current considerations and future perspectives**
JJ Boelens (UMCU, Utrecht)
- 14.05-14.25 **Neurological complications of lysosomal storage disorders**
Hannerieke van den Hout (Erasmus MC, Rotterdam)
- 14.25-14.45 **Diagnosis and follow up of MPS patients using multiplex GAG analysis**
Naomi van Vlies (AMC, Amsterdam)
- 14.45-15.00 **Ten years of the international Pompe survey**
Chris van der Meijden (Erasmus MC, Rotterdam)
- 15.10-15.55 **Vrije voordrachten**
Sessievoorzitters: Janneke Langendonk, Rebecca Heiner-Fokkema
- Progression of hip dysplasia in MPS I Hurler patients after successful hematopoietic stem cell transplantation**
Eveline Langereis (AMC, Amsterdam)
- Analysis pathogenic variants of Iduronate- 2-sulfatase in 17 Dutch patients with MPS II**
Audrey Vollebregt (Erasmus MC, Rotterdam)
- Altered interaction and distribution of glycosaminoglycans and growth factors in Mucopolysaccharidosis type I bone disease**
Sandra Kingma (AMC, Amsterdam)
- 15.55 Uitreiking prijs “Beste abstract” door het organiserend comité
- 16.00 **AFSLUITING DOOR DE VOORZITTER EN BORREL**