



## ESN najaarsvergadering donderdag 19 en vrijdag 20 november 2020

### Programma 19 november 13.55-16.25 uur

13.55 – 14.00 uur Opening door Terry Derks, voorzitter ESN

Dagvoorzitter: **Monique de Sain**

14.00 – 14.45 uur      Ethische bedenkingen rond diagnostiek en daaraan verbonden behandelingen van zeldzame ziekten  
**Prof. Dr. Martina Cornel, AmsterdamUMC**

2x Pitch

14.45 – 14.55 uur      Treatment outcome in classic-infantile Pompe disease: effects of dosing in clinical practice.  
**Imke Ditters, ErasmusMC**

14.55 – 15.05 uur      Monoamine oxidase A activity in fibroblasts as a functional confirmation of MAOA variants  
**Tessa Peters, RadboudUMC**

15.05 – 15.20 uur      *Pauze (sponsors in beeld)*

15.20 – 16.05 uur      Next generation sequencing as first tier for newborn screening: long way to go  
**Prof. Dr. Francjan J van Spronsen, UMCG**

2x Pitch

16.05 – 16.15 uur      GFUS-CDG, a new glycolysation disorder treatable with L-fucose  
**Saskia Wortmann, Paracelsus Medical University (Salzburg, Austria), RadboudUMC**

16.15 – 16.25 uur      Abnormal VLCADD newborn screening resembling MADD in four neonates with decreased riboflavin levels and/or reduced VLCAD activity  
**Marne Hagemeyer, ErasmusMC**

## Programma 20 november 13.55 – 16.20 uur

13.55 – 14.00 uur Opening door voorzitter ESN

**Dagvoorzitter: Maaïke Oosterveer**

**14.00 – 14.45 uur** Gene editing in the clinic  
**Dr. Mike Broeders, ErasmusMC**

**2x Pitch**

**14.45 – 14.55 uur** Prime editing for functional repair in patient-derived disease models  
**Imre Schene, UMC Utrecht**

**14.55 – 15.05 uur** An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids  
**Sacha Ferdinandusse, AmsterdamUMC**

**15.05 – 15.30 uur** *Pauze (sponsors in beeld)*

**15.30 – 15.50 uur** Update UMD  
**Prof. Clara van Karnebeek, RadboudUMC**  
**Prof. Hans Waterham, AmsterdamUMC**

**Pitch**

**15.50 – 16.00 uur** Intracellular nucleotide(sugar) fluxomics as a novel strategy to unravel IEMs  
**Laura Steinbusch, MUMC+**

**16.00 – 16.20 uur** Uitreiking stimuleringsbeurzen

**16.20 uur** Afsluiting door Terry Derks, voorzitter ESN