**Symposium Program**

**Venue: De Doelen**

**Rotterdam, 6-7 sept 2019**

**Friday, September 6:**

**14.30 Registration**

**Introduction session (15.30/16.00)**

**Welcome by *Gajja Salomons***

**15.30 Cerebral creatine deficiency syndromes and diagnosis**   
*Gajja Salomons/ Abel Thijs*

**15.45 Patients perspectives**   
*Xtraordinaire / ACD*

**Session 1: Signs and Symptoms of Cerebral Creatine Deficiency Syndromes (16.00/19.30)**

**16.00 Observational Study / Databases / Prevalence:**

Vigilan: Observational Study of Male CTD Patients.   
*Judith Miller*

Databases and family perspectives

*Xtraordinaire, ACD*

Prevalence in France & Netherlands

*Aurore Curie*

**16.45 The Basics of Behaviour: the Complexity of Person and Environment**

**On cognition, development and functioning.**

*Sylvia Huisman*

17.30 Break

**17.50****Importance of early diagnosis: newborn screening GAMT.**

*Marzia* *Pasquali*

**18.15 Models to improve treatment in GAMT deficiency**   
*Andreas Schulze*

**18.40 Brain magnetic resonance spectroscopy and creatine measurements.**

*Petra Pouwels*

**19.05 Q&As**

**19.45 Dinner**

**Saturday, September 7:**

8.30 coffee

**9.00 Creatine Transporter Deficiency in females**

*Jiddeke van de Kamp*

**Session 2: Animal Models for Cerebral Creatine Deficiency Syndromes (9.15/12.00)**

**9.15 Creatine deficiency mouse models (AGAT, GAMT)**

*Chi-un Choe / Arend Heerschap*

**9.40 A new knock-in rat model of creatine transporter deficiency & Gene Therapy**

*Olivier Braissant*

**10.15 CTD: New insights into epileptic phenotype.**

*Laura Baroncelli*

10.40 Coffee break

**11.00 CTD mouse model experiences and gene therapy.**

*Matthew Skelton.*

**11.25 CTD mouse model experiences and therapeutic options.**

*Ton de Grauw*

11.50 Lunch

### **Session 3: Supplementation Treatment in Cerebral Creatine Deficiency and NBS (13.00/14.00)**

**Chair: Monique Williams**

**13.00 Treatment outcomes of cerebral creatine deficiency syndromes.**   
*Saadet Andrews*

**13.20 Epilepsy in cerebral creatine deficiency syndromes.**   
*Saadet Andrews*

13.40 Q&As

14.00 Break

**Session 4: Development of New Treatment in Creatine Transporter Deficiency (14.20/19.00)**

**Chair: Olivier Braissant**

**14.20 Drug development for neurodevelopmental disorders: lessons learned from fragile X syndrome.**   
*Vincent des Portes/ Aurore Curie.*

**Clinical trials for rare disease: current pitfalls and new perspectives.**

*Vincent des Portes/ Aurore Curie.*

**15.00 Di-acetyl creatine ethyl ester therapy for creatine transporter deficiency.**    
*Maurizio Balestrino*

**15.25 Intranasal administration Dodecyl creatine ester-loaded nanoemulsion in CTD.**

*Aloise Mabondzo*

**16.50 Rescue by 4-phenylbutyrate of several misfolded creatine transporter -1 variants linked to creatine transporter deficiency syndrome.**   
*Sonja Sucic*

16.15 Panel discussion / Q&As

**Final remarks**

Drinks

18.00 end