

Program February 14th ENCORE-ITHACA Symposium

9.30-10.00 Welcome, coffee

10.00-10.10 Tjitske Kleefstra, opening

10.10-10.40 Marie Claire de Wit

Building the multi-disciplinary ENCORE expertisecentre: 12.5 years experience of doctors and families

10.40-11.10 Emma Palmer, clinical geneticist Sydney CH, Australia

Expertise clinics for neurodevelopmental disorders in Sydney and co-design of national recommendations for rare disease care in Australia

11.10-11.40 Sid Srivastava, childneurologist Boston CH, USA

Natural history studies and clinical trials as part of the Developmental Synaptopathies Consortium -Lessons Learned from Clinical Trials for Cognition and Behavior in Genetic Neurodevelopmental Disorders

11.40-12.10 Sabine Mous & Kamil Hiralal

Autism spectrum symptomatology in children with rare genetic neurodevelopmental disorders

12.10-12.40 Stefan Barakat

From aquarium tot bedside: *AMFR-dysfunction causes severe spasticity* amenable to statine treatment in a pre-clinical model

Lunch 12.40-13.30

13.30-14.10 Angela Morgan & Lottie Morison, Murdoch Children's Research Institute Melbourne, Australia

Speech-Language characterization and guidance in genetic neurodevelopmental disorders

14.10-14.40 Danielle Veenma & Geeske van Woerden

CAMK2 syndrome expertisecentre: one and one = 3

14.40-15.10 Karen Bindels de Heus & Doesjka Hagenaar

Clinical research in the Angelman expertise centre

15.10-15.40 Ype Elgersma

The need for pre-clinical research to improve care for rare genetic disorders

15.40 Tjitske Kleefstra, closure and drinks

