

PROGRAM

- 09:00 Walk-in – take own coffee, look around, solve technical issues
- 09:30 Welcome and opening
Mieke van Haelst & Terry Vrijenhoek
- 09:40 **Sandkuyl Lecture**
- Genetics delivers high precision science & medicine; can we also deliver unbiased science & medicine?
Dr. Michel Nivard, Amsterdam UMC
- 10:20 Mini-break
- 10:30 **Session 1: National initiatives for therapy development in rare diseases**
- Dutch Centre for RNA therapeutics
Prof. dr. Aartsma-Rus, LUMC
Dr. W. van Roon-Mom, LUMC
- ‘N-of-one’ for all
Prof. dr. C. van Karnebeek, Radboudumc
- Round Table discussion
- 11:15 Mini-break
- 11:20 **Session 2: Experiences with rare disease therapeutic trials**
- Targeted therapy in patients with PIK3CA related overgrowth syndromes
Prof. dr. G. Canaud, Université de Paris, France
- Clinical effect studies in Kleeftstra syndrome
Prof. dr. T. Kleeftstra, Radboudumc
- Therapeutic opportunities for obesitas
Dr. E. van den Akker, Amsterdam UMC
- Round table discussion
- 12:20 Lunch break
- 13:00 **Session 3: Current genome initiatives: regional, national, European**
- Beyond 1+ Million Genomes
Dr. R. Kok, Dutch Techcentre for Life Science
- GOALL study
Prof. J. van Meurs, Erasmus MC
- HUGE-NL
Dr. T. Vrijenhoek, UMCU
- Round table discussion
- 13:55 Mini-break
- 14:05 **Young Investigator Award**

- 14:15 **Session 4: Hot & Happening**
- Antisense oligonucleotide treatment rescues UBE3A expression and multiple phenotypes of an Angelman Syndrome mouse model
Claudia Milazzo, Erasmus MC
- Inherited variants in CHD3 demonstrate variable expressivity in Snijders Blok-Campeau syndrome
Jet van der Spek, Radboudumc
- Reduced MUNC18-1 levels, synaptic proteome changes and altered network activity in iPSC-derived neurons from six STXBP1-Syndrome patients
Annemiek van Berkel, Amsterdam UMC- locatie VU
- Identification of enhancer regions to expose novel genetic causes of spinocerebellar ataxia
Fatemeh Ghorbani, UMC Groningen
- 15:05 Mini-break
- 15:15 **Session 5: Keynote Lecture**
- Outcompeting cancer by personalized chemoprevention
PhD. Louis Vermeulen, Amsterdam UMC
- 16:00 **Closure**
Mieke van Haelst & Terry Vrijenhoek
- 16:10 **NVHG Algemene ledenvergadering (ALV)**
Aansluitend: uitreiking puzzelprijs