# Wetenschapsdag VKGN/VKGL

**16 maart 2023, In de Driehoek, Willemsplantsoen 1 C, 3511 LA Utrecht en hybride via Zoom**

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| **9.30 uur** | **Ontvangst met koffie/thee** |
| **9.55 uur** | **Welkom** |
| **10.00-12.00 uur** | **Ochtendsessie** *(voorzitter: Manon Suerink)* |
| 10.00-10.15 uur | Exploring the future prospects of a Dutch NGS-based newborn screening by investigating technical possibilities of targeted NGS, WES and WGS<br>- *Gea Kiewiet, Laboratoriumspecialist i.o. Klinische Genetica UMCG* |
| 10.15-10.30 uur | Biallelic variants in SNAPIN are associated with a novel foetal neuroanatomical phenotype<br>- *Maayke de Koning, Arts-onderzoeker Klinische Genetica LUMC* |
| 10.30-10.45 uur | De novo missense variants in RRAGC lead to a fatal mTORopathy of early childhood<br>- *Margot Reijnders, AIOS Klinische genetica ErasmusMC* |
| 10.45-11.00 uur | Obesity and loss-of-function GNB1 variants: A new form of syndromic obesity?<br>- *Lotte Kleinendorst, AIOS klinische genetica Amsterdam UMC* |
| 11.15-11.30 uur | Genome wide DNA methylation assessment in obesity genetics clinics<br>- *Niels Vos, Arts-onderzoeker Humane genetica Amsterdam UMC* |
| 11.30-11.45 uur | Effects of clonazepam in patients with ARID1B-related intellectual disability<br>- *Eline van der Sluijs, Arts-onderzoeker Klinische Genetica LUMC* |
| 11.45-12.00 uur | The power of parent driven research – 10 years Chromosome 6 Project<br>- *Aafke Engwerda, Arts-onderzoeker Afdeling Genetica UMCG* |
| **12.00-13.00 uur** | **Lunch** |
| **13.00-14.15 uur** | **Middagsessie** *(voorzitter: Margot Reijnders)* |
| 13.00-13.15 uur | Data driven criteria for genetic testing result in an efficient selection of patients with genetic dementia in a clinical setting<br>- *Sven J. van der Lee, Arts-onderzoeker Humane genetica Amsterdam UMC* |
| 13.15-13.30 uur | Genetic risk stratification in NOTCH3-associated stroke and vascular dementia<br>- *Julie Rutten, AIOS Klinische Genetica LUMC* |
| 13.30-13.45 uur | Biallelic NDC1 variants that interfere with ALADIN binding in neuropathy and Triple-A-like syndrome<br>- *Jordy Dekker, PhD-student Klinische genetica ErasmusMC* |

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| 13.45-14.00 uur | AMFR dysfunction causes spastic paraplegia amenable to statin treatment in a preclinical model<br>- *Stefan Barakat, AIOS Klinische genetica ErasmusMC* |
| 14.00-14.15 uur | Genetic causes of stillbirth<br>- *Manon Suerink, AIOS klinische genetica LUMC* |
| **14.15-14.45 uur** | **Thee- en koffiepauze** |
| **14.45-16.00 uur** | **Middagsessie** *(voorzitter: Mala Misra-Isrie)* |
| 14.45-15.00 uur | Energetic compensation prevents phenotype development in carriers of a truncating TTN variant<br>- Job Verdonschot, AIOS klinische genetica MUMC+ |
| 15.00-15.15 uur | Clinically-relevant Germline Variants are Common in Children with Non-Medullary Thyroid Cancer<br>- *Karin van der Tuin, AIOS klinische genetica LUMC* |
| 15.15-15.30 uur | Cancer predisposition genepanel sequencing among all children with cancer – Yes, we can, but should we?<br>- *Jette J. Bakhuizen, AIOS klinische genetica UMC Utrecht* |
| **15.30-16.00 uur** | **Juryberaad en borrel** |
| **16.00-17.00 uur** | **Prijsuitreiking en vervolg borrel** |