

Molecular Medicine
Postgraduate School



SOPHIA
GENETICS

Programme

NGS course: Genomic Resequencing in Medical Diagnostics 2015

Date: Tuesday 22nd - Thursday 24th September
Venue: Onderwijscentrum, Erasmus MC, Rotterdam

VKGL & VKGN accreditation (tbc.): 6 points per day

Jasper Saris, Christian Gilissen, Rob van der Luijt, Johan den Dunnen

Fourth Edition of Three Day Course on Variant Detection and Interpretation in a Diagnostic Context

vs. 150918'

The Book of Life... how to read it, what do you read, and what does it actually mean?

Introduction

VKGL/VKGN 'NGS in DNA diagnostics' is a course aimed at Genomic Resequencing in a Medical Diagnostic Context, i.e. to apply Next Generation Sequencing data as diagnostic tool in the hospital. Lectures will be accompanied with hands-on exploration.

Program Outline

- Day 1 consists of an introduction to NGS techniques followed by specialised lectures on sample preparation, enrichment technologies and read mapping.
- Day 2 will continue with lectures on variant calling, annotation and interpretation with a diagnostic emphasis applied to Gene Capturing Panels, Exome Screening and CNV analysis.
- The afternoon of Day 1 & Day 2 is reserved for workshops and software demonstrations. Real data will be used where possible in hands-on tutorials using both commercial and open source software.
- Day 3 focuses on impact of results in the lab and ethical and legal issues of NGS in the clinical application. Real-life applications of NGS in the clinic will be presented. A forum discussion between researchers and clinicians about mutual expectations, ethics, implications of NGS on diagnostics and data sharing concludes the day.

Target audience

This course is intended for bioinformaticians, researchers and molecular & clinical geneticists. The mixed programme will appeal to all those requiring a solid background in the application of NGS techniques for (routine) genetic diagnosis of a patient. The course will be mainly in English.

Organisation, Accreditation and Registration

The course is a joint effort from individuals of the NVHG/VKGL/VKGN (diagnostic) community of The Netherlands. Accreditation is be filed for at VKGL and VKGN (6 points; GAIA registration is pending).

ECTS

This course including self-study is 1,0 ECTS.

Date and Location

The course will be held at the Erasmus Medical Centre, Medical Faculty, at the 'Onderwijscentrum' (Educational Centre, rooms are mentioned 'OWR no. NN').

- The course will start in lecture room no. 35.
- Additional rooms will be used depending on the programme.

For maps of the Erasmus MC, including the Educational Centre, see: www.molmed.nl


Day 1 - Tuesday 22nd; NGS-based testing

(somewhat aimed at: Lab Specialists & Bioinformaticians)

| Time | Title | Speaker (center) | Location |
|------------------------------------|--|----------------------------------|----------|
| Moderator: Johan den Dunnen | | | |
| 09.00-9.30 | Coffee & Registration | | OWR-35 |
| 9.30-10.00 | Characteristics of Sequencing Methods | Johan den Dunnen (LUMC) | OWR-35 |
| 10.00-10.45 | Enrichment Techniques, Amplified panel / exome | Wilfred van IJcken (Erasmus MC) | OWR-35 |
| 10.45-11.15 | Coffee break | | OWR-35 |
| 11.15-12.00 | Mapping sequence reads & Calling variants | Robert Kraaij (Erasmus MC) | OWR-35 |
| 12.00-12.30 | WES or WGS... statistics & power in practice | Gijs Santen (LUMC) | OWR-35 |
| 12.30-13.15 | Lunch | | OWR-35 |
| 13.15-14.00 | Experiment setup, QC & SNP profiling panel for sample tracking | Chris Mattocks (NGRL Wessex, UK) | OWR-35 |
| 14.00-14.30 | Annotation of variants | Joeri van der Velde (UMCG) | OWR-35 |
| 14.30-14.40 | <i>Beauty Contest (3x3 min)</i> Parallel workshops Introduction (30 min) & workshop (90 min) | NextGene, Qiagen, hAwk | OWR-35 |
| 14.40-15.00 | Coffee Break + Relocation | | OWR-35 |
| Parallel S1 15.00-17.00 | NextGene (Softgenetics, Bioké) | Fons Elstrodt | OWR-22 |
| Parallel S2 15.00-17.00 | Qiagen Clinical Insight (aka Ingenuity Clinical) | Ian Cook | Ee-15.28 |
| Parallel S3 15.00-17.00 | hAwk, Genepanel QC and SNV Calling | Walter de Valk & Frank Sleutels | COO-6 |

Day 2 - Wednesday 23th; Data analysis and variant interpretation

(aimed at: lab specialists and Genetic Counselors)

| Time | Title | Speaker | Location |
|--------------------------------|--|--|---|
| Moderator: Jasper Saris | | | |
| 9.00 - 9.45 | CNV calling in Gene Panels, Exomes and WGS | Thomas Kuilman (NKI) | OWR-35 |
| 9.45 - 10.15 | Individual Variant Interpretation | Nienke vd Stoep (LUMC) | OWR-35 |
| 10.15 - 10.45 | Coffee break | | OWR-35 |
| 10.45 - 11.15 | Gene Panel Experience | Mariëlle van Gijn (UMCU) | OWR-35 |
| 11.15 - 11.45 | Tiered WES or Exome masking (icm LOVD+) | Claudia Ruivenkamp (LUMC) | OWR-35 |
| 11.45 - 12.15 | NIPT - is trisomy screening the lower limit? Is 2Mb resolution feasible? | Erik Sistermans (VUMC) | OWR-35 |
| 12.15 - 13.15 | Lunch | | OWR-35 |
| 13.15 - 13.45 | Somatic Mutation Detection in Cancer | Maartje Vogel (NKI) | OWR-35 |
| 13.45 - 14.15 | HT-Metabolomics | Ron Wevers (UMCRadboud) | OWR-35 |
| 14.15 - 14.35 | <i>Beauty Contest (4x3 min)</i> Parallel workshops Introduction (30 min) & workshop (90 min) | Sophia Genetics, CLC Bio, Galaxy | OWR-35 |
| 14.35-14.45 | Coffee Break + Relocation | | OWR-35 |
| Parallel S1 14.45-17.00 | Sophia Genetics | Verena Tischler & Koenraad Eycken | OWR-22 |
| Parallel S2 14.45-17.00 | Qiagen Biomedical Workbench (aka CLC-bio) | Patrick Dekker & Sara Nema | Ee-15.28 |
| Parallel S3 14.45-17.00 | <i>GalaXy, free (targeted-) WES and RNAseq pipeline for single and automated analysis</i> | Saskia Hiltman, Frank Sleutels, Andrew Stubbs ea | COO-6 |
| 18.00 - ... | Dinner for all participants & speakers at: Z&M Restaurant Veerhaven 13, 3016 CJ Rotterdam, T. 010-2800980, http://www.zenrestaurant.nl/ | | |
| | <i>sponsored by Sophia Genetics</i> | |  |

Day 3 Thursday 24th; NGS - results interpretation and counseling issues

(aimed at: lab specialists and Genetic Counselors)

| Time | Title | Speaker | Location |
|--------------------------------------|--|---|----------|
| Moderator: Christian Gilissen | | | |
| 9.00-9.30 | Eurogentest guidelines (and AMCG guidelines update) in WES and WGS | Stefan White (LUMC) | OWR-35 |
| 9.30-10.15 | Whole Genome Sequencing in the clinic | Christian Gilissen (UMCRadboud) | OWR-35 |
| 10.15-10.45 | Medical Ethics & Genomics Current Legal framework en FAQ (Dutch) | Eline Bunnik (ErasmusMC) | OWR-35 |
| 10.45-11.15 | Coffee break | | OWR-35 |
| 11.15-11.45 | Informed Consent (<i>What can I tell the doctor?</i>) & Incidental Findings (<i>What can we tell the patient?</i>) | Janneke Schuurs-Hoeimakers (UMCRadboud) | OWR-35 |
| 11.45-12.30 | Active Clinical Collaboration on Test Result Interpretation... (when) Needed? | Gijs Santen (LUMC) | OWR-35 |
| 12.30-13.30 | Lunch | | OWR-35 |
| 13.30-14.15 | WGS for Neonates in trouble | Cleo van Diemen (UMCG) | OWR-35 |
| 14.15-15.00 | Functional Validation of Variants in a Diagnostic Context | Frans Verheijen (Erasmus MC) | OWR-35 |
| 15.00-15.30 | Coffee break | | OWR-35 |
| 15.30-16.30 | Panel Discussion - one year later (<i>with voting!</i>) | Jasper Saris + Terry Vrijenhoek | OWR-35 |
| 16.30-17.00 | Return badges & Hand in evaluation forms | | OWR-35 |

Registration fees

- €250 for PhD students (€200 for 2 days)
- €500 for researchers at universities or non-profit research institutes (€400 for 2 days)
- €1000 for researchers from profit organizations (industry) (€800 for 2 days)

Additional remark: AIOs are not considered PhD-students and will thus also not receive the PhD-discount.

PAYMENT: MGC Notification

The Erasmus/LUMC Research School MGC has made a selection of courses it deems fit for its members. This course is not one of them.

Therefore, the Erasmus/LUMC Research School MGC will not pay for its members to participate in this course. All MGC-members will need to pay themselves (or from personal budget) after receiving an invoice (please provide personal address) or will have their research departments pay for them (in that case, please provide 'kostenplaatsnummer' of the research group). To see if you are an MGC-member, here you can find an overview of all MGC research groups: www.medgencentre.nl

Invoice: Fees can be paid upon an INVOICE. After your registration you will receive the INVOICE per post.

Cancellations

The fees are for all the days of the Course. There is no discount for partaking only in part of the Course. Our cancellation policy is that cancellation is possible up to one week before the start of the Course. Later cancellation will not be accepted, but you are allowed to send a substitute.