

Programme

NGS course: Genomic Resequencing in Medical Diagnostics 2014

Date: Tuesday 28th - Thursday 30th October

Venue: Onderwijscentrum, Erasmus MC, Rotterdam

VKGL & (applied for) VKGN accreditation: 6 points per day

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

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

vs. 141027

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 nr. NN').

- The course will start in lecture room no. 36.
- Additional rooms will be used depending on the programme.


See for maps of the Erasmus MC, including the Educational Centre, the MolMed website: www.molmed.nl

Day 1 - Tuesday 28th; NGS-based testing

(aimed at: Lab Specialists & Bioinformaticians)

| Time | Title | Speaker (center) | Location |
|------------------------------------|--|--|----------|
| Moderator: Johan den Dunnen | | | |
| 09.00-9.30 | Coffee & Registration | | OWR-36 |
| 9.30-10.00 | Characteristics of Sequencing Methods | Johan den Dunnen (LUMC) | OWR-36 |
| 10.00-10.45 | Enrichment Techniques, Amplified panel / exome | Wilfred van Ijcken (Erasmus MC) | OWR-36 |
| 10.45-11.15 | Coffee break | | OWR-36 |
| 11.15-12.00 | Mapping sequence reads & Calling variants | Laurent Francioli (UMCU) | OWR-36 |
| 12.00-12.30 | WES or WGS... statistics & power in practice | Gijs Santen (LUMC) | OWR-36 |
| 12.15-13.15 | Lunch | | OWR-36 |
| 13.15-14.00 | Experiment setup, QC & SNP profiling panel for sample tracking | Chris Mattocks (NGRL Wessex, UK) | OWR-36 |
| 14.00-14.30 | Annotation of variants | Joeri van der Velde/ Morris Swertz (UMCG) | OWR-36 |
| 14.30-15.00 | Coffee break | | OWR-36 |
| 14.50-15.00 | <i>Beauty Contest (3x3 min)</i> Parallel workshops Introduction (30 min) & workshop (90 min) | NextGene, Ingenuity | OWR-36 |
| Parallel S1 15.00-17.00 | NextGene (Softgenetics, Bioké) | Fons Elstrodt | COO-6 |
| Parallel S2 15.00-17.00 | Ingenuity (Qiagen) | Arne Arens | Ee-15.28 |
| Parallel S3 15.00-17.00 | Giving consent in 2028 | Terry Vrijenhoek | OWR-36 |

Day 2 - Wednesday 29th; 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 | Victor Guryev (UMCG) | OWR-36 |
| 9.45 - 10.15 | Individual Variant Interpretation | Nienke vd Stoep (LUMC) | OWR-36 |
| 10.15 - 10.45 | Coffee break | | OWR-36 |
| 10.45 - 11.15 | Cardio Gene Panel Experience | Jan Jongbloed (UMCG) | OWR-36 |
| 11.15 - 11.45 | Multiple Gene Panel approach vs Exome masking | Marjon van Slegtenhorst | OWR-36 |
| 11.45 - 12.15 | NIPT - is trisomy screening the lower limit? | Elles Boon (LUMC) | OWR-36 |
| 12.15 - 13.15 | Lunch | | OWR-36 |
| 13.15 - 13.45 | Somatic mutation detection in Cancer | Marco Koudijs (UMCU) | OWR-36 |
| 13.45 - 14.15 | HT-metabolomics | Ron Wevers (UMCRadboud) | OWR-36 |
| 14.15 - 14.35 | Coffee break | | OWR-36 |
| 14.35-14.45 | <i>Beauty Contest (4x3 min)</i> Parallel workshops Introduction (30 min) & workshop (90 min) | Sophia Genetics, Cartagenia, CLC Bio, hAwk | OWR-36 |
| Parallel S1 14.45-17.00 | Sophia Genetics | Zhenyu Xu | OWR-22 |
| Parallel S2 14.45-17.00 | Cartagenia | Ewoud Ouwerkerk & Jonatan Taminou | OWR-36 Laptops necessary |
| Parallel S3 14.45-17.00 | CLC Bio | Patrick Dekker | Ee-15.28 |
| Parallel S4 14.45-17.00 | hAwk | Walter de Valk & Frank Sleutels | Ee-24.34 |
| 18.00 - ... | <i>Dinner for all participants & speakers at: Z&M Restaurant Veerhaven 13, 3016 CJ Rotterdam, T. 010-2800980 sponsored by Sophia Genetics</i> | |  |

Day 3 Thursday 30th; NGS - results interpretation and counseling issues

(aimed at: lab specialists and Genetic Counselors)

| Time | Title | Speaker | Location |
|--|---|---------------------------------|----------|
| Moderator: Christiaan Gillissen | | | |
| 9.00-9.30 | Eurogentest guidelines (and AMCG guidelines update) | Gert Matthijs (KU Leuven) | OWR-36 |
| 9.30-10.00 | Informed Consent; What can I tell the doctor? & Incidental Findings; What can I tell the patient? | Dorien Lugtenberg (UMCN) | OWR-36 |
| 10.00-10.30 | Medical Ethics & Genomics Current Legal framework en FAQ (Dutch) | Corrette Ploem (AMC) | OWR-36 |
| 10.30-11.00 | Coffee break | | OWR-36 |
| 11.00-11.45 | Whole Genome Sequencing in the clinic | Christian Gilissen (UMCRadboud) | OWR-36 |
| 11.45-12.30 | WGS for Neonates in trouble | Jan Jongbloed (UMCG) | OWR-36 |
| 12.30-13.30 | Lunch | | OWR-36 |
| 13.30-14.15 | Using HPO fenotyping & Databases | Rolf Sijmons (UMCG) | OWR-36 |
| 14.15-15.00 | Functional validation of variants in a diagnostic context | Frans Verheijen (Erasmus MC) | OWR-36 |
| 15.00-15.30 | Coffee break | | OWR-36 |
| 15.30-16.30 | Panel Discussion - one year later <i>(with voting!)</i> | Jasper Saris (Erasmus MC) | OWR-36 |
| 16.30-17.00 | Return badges & Hand in evaluation forms | | OWR-36 |

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)
- Participants working in a CGD-project (Centre for Genome Diagnostics) receive a 25% discount on the relevant fee.

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.