Programme
NGS course: Genomic Resequencing in Medical Diagnostics 2018

Date: Tuesday 4th – Thursday 6th September
Venue: Onderwijscentrum, Erasmus MC, Rotterdam

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

Jasper Saris, Christian Gilissen, Johan den Dunnen

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

vs. 180704

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 medical practice.
Lectures will be accompanied with hands-on exploration of data and software.

Program Outline
- Day 1 consists of an introduction to NGS techniques followed by specialized 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 some CNV analysis.
- The afternoons of Day 1 & Day 2 are 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 molecular and clinical geneticists, and bioinformaticians and researchers in the field and to some extent pathologists. 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.

Organization, Accreditation and Registration
The course is a joint effort from individuals of the NVHG/VKGL/VKGN (diagnostic) community of The Netherlands. Accreditation is 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, 'Onderwijscentrum' (i.e. Educational Centre, rooms are mentioned 'OWR no. NN').

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

For maps of the Erasmus MC, including the Educational Centre, see: www.molmed.nl
In short: From Metro Dijkzigt enter via Sophia Hospital, turn right, proceed right on to and climb white staircase, enter hall of OWR, take yellow stairs to level 1 of OWR.

Day 1 – Tuesday 4th; NGS-based testing --Preliminary

(somewhat aimed at: Lab Specialists & Bioinformaticians)

Time	Title	Speaker (center)	Location
Moderator: Jasper Saris			
9.00-9.30	**Coffee & Registration**		CQ-F1
9.30-10.15	Characteristics of Sequencing Methods	Johan den Dunnen (LUMC)	CQ-F1
10.15-11.00	Enrichment Techniques, Amplify or Capture	TBC	CQ-F1
11.00-11.15	**Coffee break**		CQ-F1
11.15-12.00	Mapping sequence reads & Calling variants	Martin Elferink (UMCU)	CQ-F1
12.00-12.30	Experiment setup, QC & sample tracking	Koen van Gassen (UMCU)	CQ-F1
12.30-13.15	**Lunch**		CQ-F1
13.15-14.00	Annotation of variants	Joeri van der Velde (UMCG)	CQ-F1
14.00-14.30	WES or WGS… statistics & power in practice	Gijs Santen (LUMC)	CQ-F1
14.30-17.00	*Beauty Contest (3x3 min)* Coffee Break 2 Parallel workshops *Introduction (30 min) & workshop (90 min)*		CQ-F1
14.40-15.00	**Coffee Break + Relocation**		CQ-F1
Parallel S1 15.00-17.00	Cartagenia – Bench NGS - Variant Assessment	TBC	CQ-F1
Parallel S2 15.00-17.00	WES Pedigree Analysis using Galaxy	Andrew Stubss (Erasmus MC)	Ee-15.28

Day 2 - Wednesday 5th[th]; Data analysis and variant interpretation - -Preliminary

(aimed at: lab specialists and Genetic Counselors)

Time	Title	Speaker	Location
Moderator: Jasper Saris			
9.30 - 10.15	CNV calling in Gene Panels, Exomes and WGS	Jayne Hehir-Kwa (PMC)	CQ-F1
10.15 – 11.00	Individual Variant Interpretation	Nienke vd Stoep (LUMC)	CQ-F1
11.00 - 11.15	**Coffee break**		CQ-F1
11.15 - 11.45	Tiered WES or Exome masking (icm LOVD+)	Claudia Ruivenkamp (LUMC)	CQ-F1
11.45 - 12.30	NIPT – just fetal trisomy screening or more?	Diane van Opstal (Erasmus MC)	CQ-F1
12.30 - 13.15	**Lunch**		CQ-F1
13.15 - 13.45	Somatic Mutation Detection in Cancer	Maartje Vogel (AvLI)	CQ-F1
13.45 – 14.15	HT-Metabolomics	TBC	CQ-F1
14.15 – 17.00	*Beauty Contest (3x3 min)* Coffee Break 2 or 3 Parallel workshops *Introduction (30 min) & workshop (90 min)*		CQ-F1
14.35 - 14.45	**Coffee Break + Relocation**		CQ-F1
Parallel S1 14.45 - 17.00	*LOVD3+* Variant Prioritisation, Classification and Interpretation	Claudia Ruivenkamp & Ivo Fokkema (LUMC)	Ee-15.28
Parallel S2 14.45 - 17.00	Ingenuity Variant Analysis / Clinical Insight	TBC	C00-3
Parallel S3 14.45 - 17.00	TBC	TBC	CQ-F1
18.00 - …	***Dinner for all participants & speakers at: Z&M Restaurant.*** ***Address: Veerhaven 12-13, 3016 CJ Rotterdam Tel: 010 280 0980***		

Day 3 Thursday 6th; NGS – results interpretation and counseling issues -- Preliminary

(aimed at: lab specialists and Genetic Counselors)

Time	Title	Speaker	Location
Moderator: Christian Gilissen			
9.30 - 10.15	Whole Genome Sequencing in the clinic	Christian Gilissen (UMC Radboud)	CQ-F1
10.15 - 10.50	Medical Genomics Current Legal framework & FAQ (Dutch)	Eline Bunnik (Erasmus MC)	CQ-F1
10.50 - 11.10	**Coffee break**		CQ-F1
11.10 - 11.45	Informed Consent (*What can I tell the doctor?*) & Incidental Findings (*What can we tell the patient?*)	Helger Yntema (Radboud UMC)	CQ-F1 CQ-F1
11.45 - 12.30	Active Clinical & Lab Collaboration on Test Result Interpretation and use of HPO	Gijs Santen (LUMC)	CQ-F1
12.30 - 13.15	**Lunch**		CQ-F1
13.15 – 14.00	WGS & HPO for Neonates in Trouble	Cleo v. Diemen & Mieke Kerstjens (UMCG)	CQ-F1
14.00 - 14.30	Clinical use of RNA seq	TBC	CQ-F1
14.30 - 15.00	Functional Validation of Variants in a Diagnostic Context	Frans Verheijen (Erasmus MC)	CQ-F1
15.00 - 15.20	**Coffee break**		CQ-F1
15.20 - 16.15	Panel Discussion – one year later *(with voting!)*	Jasper Saris + Terry Vrijenhoek	CQ-F1
16.15 – 16.30	**Return badges & Hand in evaluation forms**		CQ-F1