NGS data analysis workshop @ Nijmegen-NL

Details:

- October 9th 2018.
- @Nijmegen, The Netherlands.
- Registration via: https://biotexcel.com/event/genomic-medicine-2018-Nijmegen/#registration.
- In combination with **Genomic Medicine 2018 Nijmegen** conference (October 10 & 11 2018)!

Program Workshop-October 9th:

- Introduction to NGS.
- Introduction to the workshop.
- Workshop NGS Bioinformatic basics.
- Workshop NGS Variant interpretation.
- Workshop NGS Diagnostic interpretation.

Information on the NGS data analysis workshop:

- Workshop NGS Bioinformatic basics: basic data formats and analysis steps for analyzing NGS data from exome and whole genome data.
- Workshop NGS Variant interpretation: interpretation of the thousands of variants identified in an NGS experiment to identify the cause of a genetic disease.
- Workshop NGS Diagnostic interpretation: working with a diagnostic sample and learning how to interpret exome data from a diagnostic perspective.

Workshop organizers:

- **Genome Diagnostics Nijmegen, Radboud University Medical Center**
- **Eurogentest**
- **Biotexcel**

Information on the instructors

Dr. Christian Gilissen

Christian Gilissen has a master's degree in computer science and works as a post-doc in bioinformatics within the Translational Genomics team. He is specialized in the analysis of Next Generation Sequencing data and interpretation of single nucleotide variation. In 2012 he was awarded the Young Investigator Award by the Dutch bioinformatics consortium for his thesis on the identification of disease genes through next generation sequencing methods. In 2013 he obtained a Dutch Veni grant to study bioinformatics methods to interpret single nucleotide variants in relation to disease. He is actively involved in the implementation of bioinformatics methods, infrastructure and automation to incorporate new technologies in diagnostics in order to improve the routine diagnostic process.

Jordi Corominas Galbany

Jordi Corominas Galbany studied biotechnology at the Autonomous University of Barcelona after which he started his PhD in 2009 at the Centre for Research in Agricultural Genomics under the supervision of Dr. Josep Maria Folch Albareda and Dr. Maria Ballester Devis. During his PhD he worked for 8 months in the Animal Genetic and Integrative Biology group at the French National Institute for Agricultural Research (INRA) in Jouy-en-Josas (France), under the supervision of Dr. Emmanuelle Bourneuf and Dr. Jordi Estellé. In this time, he worked on the identification of copy number variants in a porcine model of cutaneous melanoma (MeLiM pigs). He obtained his PhD in 2013 on "Functional genomics and candidate genes for meat quality traits in pigs", for which he received the Extraordinary doctorate award.

After his PhD he started as a post-doctoral researcher at the department of Human Genetics of the Radboud university medical center in Nijmegen. His research was focused on the use of whole exome sequencing data from families and big cohorts to identify novel disease-causing genes/variants associated with multi-factorial disorders: age-related macular degeneration and attention-deficit/hyperactivity disorder. In 2017 he started working as a bioinformatician at Genome Diagnostics Nijmegen, where he is focused on the development and optimization of pipelines for structural variants calling in next-generation sequencing data.

Maartje van der Vorst

Maartje van de Vorst finished her Bachelor of Applied Science at the Avans University of Applied Sciences Breda in 2013 with a specialization in biotechnology and bioinformatics. After her graduation she started working as a bioinformatician at the department of Human Genetics of the Radboud university medical center in Nijmegen the Netherlands, under the supervision of Prof. Joris Veltman and Dr. Christian Gilissen. At that time she focused mainly on the interpretation of single nucleotide variants from whole genome sequencing data to identify novel genetic causes of severe intellectual disability that was published in Nature in 2014. Then she continued her work as part of the Translational Genomics group, with Dr. Lisenka Vissers and Dr. Christian Gilissen, and developed automated pipelines for the analysis of exome data and Molecular Inversion Probes (MIPs).

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