

MEDGEN

Developing Medical Genetics Education Through Curriculum Reforms
And Establishment Of Postgraduate Training Programs

11-14 April, 2016

Medical University of Graz, Austria
Training on NGS Bioinformatics Analysis
at Yerevan State Medical University, Armenia

Day 1:

1. Bioinformatics Introduction (Theory)
2. NGS Technology (Theory)
3. Data formats (Theory)
4. Quality Control (Theory and Exercise)

Day 2:

5. Variant Calling Overview (Theory and Exercise)
6. Visualization and Quality Control (Exercise)
7. Variant annotation (Theory and Exercise)
8. Variant interpretation (Theory and Exercise)

Day 3:

9. Exome sequencing (Theory and Exercise)
 - 9.1. SRA intro and data download
 - 9.2. Variant Calling
 - 9.3. Variant Filtering
 - 9.4. Variant interpretation
 - 9.5. Trio/IBD2/HomozygosityMapper
10. Human Genome in Bioinformatics (Theory)
11. CNV calling (Theory and Exercise)
12. Structural Variant Calling (Theory and Exercise)

Day 4:

13. BCBio as Pipeline Tool (Theory and Exercise)
 - 13.1. Setup
 - 13.2. SNP-calling
14. Alternative experimental setups (Theory)
 - 14.1. RNA-Seq
 - 14.2. ChIP-Seq
15. Research overview Institute of Human Genetics Graz (if wanted and time left)
16. Routine medical genetics at the Institute of Human Genetics Graz (if wanted and time left)
17. Summary, Wrap up, Questions