

# Identifying Genes for Mendelian Traits using Next Generation Sequence Data

September 28-Oct 2, 2015

Max Delbrück Center for Molecular Medicine–Berlin, Germany

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Each session will begin with a theoretical introduction followed by practical exercises. The course instructors are Suzanne Leal (Baylor College of Medicine) and Michael Nothnagel (University of Cologne).

The course will be held daily from 9:00 a.m. to 5:00 p.m., except for Wednesday, September 30<sup>th</sup> when the course will end at 1:00 p.m. in order to have free time for sightseeing. On Monday, September 28<sup>th</sup>, registration will be held from 8:30 to 9:00 a.m. A wine and cheese get together will be held for students and faculty directly after the course on September 28<sup>th</sup>.

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| <b>MONDAY</b><br>September 28 <sup>th</sup>  | Morning     | <i>Lecture</i><br>Introduction to discovering causal variants using filtering approaches and linkage analysis. Basic terminology; introduction to calculating LOD scores<br><i>Pencil and Paper Exercises</i><br>Calculation of LOD scores   |
|  | Afternoon   | <i>Lecture</i><br>Getting started using LINUX/UNIX; introduction to linkage analysis; Linkage analysis programs file formats; locus types and data entry for an autosomal dominant disease<br><i>Computer Exercises</i><br>LINKAGE/FASTLINK program (MLINK)  |
|  | 17:00-18:30 | Wine and Cheese Party  |
| <b>TUESDAY</b><br>September 29 <sup>th</sup> | Morning     | <i>Lecture</i><br>Incomplete penetrance; penetrance for autosomal recessive and x-linked inheritance; allele frequency estimation; marriage and consanguinity loops; quality control for genotype data, detecting genotyping errors and multipoint linkage analysis<br><i>Computer Exercises</i><br>MERLIN, UNKNOWN, PEDMANAGER and PEDCHECK |
|  | Afternoon   | <i>Lecture</i><br>Genetic maps; analysis under linkage admixture; overview of programs for analyzing family data; haplotype reconstruction, Homozygosity mapping, designing a family based study and who to ascertain<br><i>Exercises</i><br>GENEHUNTER, Homozygosity Mapper   |

**WEDNESDAY**  
September 30<sup>th</sup>

Morning *Lecture*  
Generation of Next Generation Sequence (NGS) data, VCF file format,  
Variant Annotation,  
Visualization of NGS data  
*Computer Exercise*  
Integrative Genome Viewer (IGV)

Afternoon Free for sightseeing

**THURSDAY**  
October 1<sup>st</sup>

Morning *Lecture*  
Quality control of NGS data  
Detecting of variants for Mendelian traits (autosomal dominant and  
recessive and x-linked) and de novo events, using filtering approaches.  
*Computer Exercises*  
*GEMINI*

Afternoon *Lecture*  
Detecting of variants using filtering approaches (continued). Performing  
homozygosity mapping using NGS data  
*Computer Exercises*  
Variant Mendelian Tools (VMT), Homozygosity Mapper

**FRIDAY**  
October 2<sup>nd</sup>

Morning *Lecture*  
Linkage analysis using NGS data, Selection of family members for NGS,  
Determining pedigree informativeness, Power analysis  
Exercise  
SEQLinkage, SLINK, MSIM

Afternoon *Lecture*  
Predicting functionality of variants using bioinformatics tools, Follow-up  
functional studies  
*Computer Exercises*  
CADD, FATHMM, GERP, LRT, Mutalyzer, Mutation Assessor, Mutation  
Taster, PhlyoP, Polyphen-2, PhastCons, PROVEAN, SIFT