Identifying Genes for Mendelian Traits using Next Generation Sequence Data

November 11-15, 2019

Max Delbrück Center for Molecular Medicine–Berlin, Germany

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, when the course will end at 1:00 pm in order to have free time for sightseeing. On Monday, registration will be held from 8:30 to 9:00 am. A wine and cheese get together will be held for students and faculty directly after the course on Monday.

MONDAY November 11th	Morning	*Lecture* Introduction to discovering causal variants using filtering approaches and linkage analysis. Basic terminology; introduction to calculating LOD scores *Pencil and Paper Exercises* Calculation of LOD scores
	Afternoon	*Lecture* Getting started using LINUX/UNIX; Cloud Computing; introduction to linkage analysis; Linkage analysis programs file formats; locus types and data entry for an autosomal dominant disease *Computer Exercises* LINKAGE/FASTLINK program (MLINK)
	17:00-18:30	Wine and Cheese Party
TUESDAY November 12th	Morning	*Lecture* 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 *Computer Exercises* MERLIN, UNKNOWN, PEDMANAGER and PEDCHECK
	Afternoon	*Lecture* 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 *Exercises* GENEHUNTER, Homozygosity Mapper

WEDNESDAY November 13th	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 November 14th	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 November 15th	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