Title: Development of a web interface for deep phenotyping of rare genetic diseases

Lecturer: Robin Steinhaus

Maximum number of participants: 3

Period/preliminary appointment: 19.04.2021 - 11.06.2021

Location: Charité/BIH - Bioinformatics & Translational Genetics, Invalidenstr. 97

Short description of content:

Precision medicine requires precise phenotypes. The correct characterisation of the symptoms of rare disorders is of utmost importance for the diagnosis, understanding and treatment of the disease. With SAMS (Symptom Annotation Made Simple), the BIH/Charité group for Bioinformatics and Translational Genetics has developed a free and simple tool for tracking symptoms in genetic diseases based on four widely used annotation systems: HPO, OMIM, Orphanet and DIMDI Alpha-IDs. SAMS has simple and intuitive web-based interfaces that allow clinicians and patients to annotate symptoms with ease. The tool will use machine learning to facilitate differential diagnosis. We are looking for up to three students to support the ongoing development of SAMS, including improving machine learning algorithms for differential diagnosis, web development and internationalisation.

Biological topics:

- Rare genetic diseases
- Annotation of symptoms and medical diagnosis
- Human Phenotype Ontology (HPO)
- Online Mendelian Inheritance in Man (OMIM)
- Orphanet

Informatics learning goals:

- Modern web development
- Client-server architecture
- Internationalisation and localisation
- Machine learning algorithms for differential diagnosis

Quantitative allocation (in %):

Practical programming work:	80%
Soft skills:	20%

Programming language(s) used: JavaScript, CSS, Terminal, SQL, Perl

Difficulty level:

Programming:	5*
Biology/Chemistry:	1*
Projekt management:	2*

Required previous knowledge:

- Some experience in the programming languages listed above
- Interest in the medical field

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