Detection of RNA alternative splicing for oncology

RNA-seq, a technology widely used in research, is now making its way into clinical practice and diagnostic laboratories. The technology allows RNA expression to be quantified but can also be used to detect structural anomalies of RNA such as gene fusions or alternative splicing. The role of gene fusions in cancer is well established and some hospitals, such as HUG, have already integrated gene fusions detection from RNA-seq data in their routine diagnostic toolkit. On the other hand, the role of alternative splicing and other RNA structure alterations in cancer is well known but has yet to be incorporated into the clinic.

The project is two-fold. First the student will perform a literature review and identify RNA structure alterations (alternative splicing, alternative start/stop or polyA site, etc.) that are relevant for oncology, i.e. that have an impact on diagnostic, prognostic or treatment options. Collaboration with the oncology department is likely. This knowledge is to be summarized into a web database such as CIViC or another system to be constructed. The second part of the project is to develop an analysis pipeline for a reliable detection of the alterations present in the database of tumor samples sequenced with a targeted RNA-seq panel. This part involves the evaluation of existing tools and the design of a novel method if deemed necessary.

The student will work under the supervision of Prof. Thomas McKee, head of the molecular pathology unit in HUG and professor in the faculty of medecine of the University of Geneva, and Dr. Yann Christinat, bioinformatician in the aforementionned unit.

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