Abstract for the keynote presentation

"In Canada, about 1500 pediatric cancers are diagnosed each year. Despite improvements in risk-based treatment protocols, ~20% of childhood cancer patients do not respond to current therapies and ultimately succumb to their disease, urging the need for new and more effective therapeutic approaches. Since individual tumours of the same clinical type harbour diverse sets of genomic alterations that drive oncogenesis and modulate drug response, personalized targeted therapy based on next generation sequencing may be a key to increase curerates and decrease treatment-related morbidity and mortality.

In this project, we implemented an automated pipeline to identify single nucleotide variants, indels, gene fusions, and copy number variations from DNA and RNA sequencing data. We use Perl wrappers to encapsulate well-established softwares (e.g. Bowtie2, STAR, PicardTools) and connect them using a common interface. Our goal is to detect prognostic markers and drug-actionable targets in a rapid and standardized way, going from biopsy to detailed tumour analysis within a clinically-relevant timeframe. Actionable variations are validated by re-sequencing or qPCR, detailed in a report with supportive information and communicated to the physician. Preliminary results on 10 cases indicate the feasibility and great potential of our approach; indeed, the final report can be in the treating oncologist's hands in less than 8 weeks. This represents a first step towards the implementation of a targeted therapy program for children with relapse or refractory cancer."