Mark Trinder, a graduate student in the MD/PhD program at the University of British Columbia, is passionate about the under-diagnosis and subsequent under-treatment of hypercholesterolemia. His approach to correcting this issue is through working to identify individuals with familial hypercholesterolemia, a significant risk factor for premature coronary artery disease. Mark works alongside his program supervisor, Dr. Liam Brunham, at the Healthy Heart Program Prevention Clinic at St. Paul’s Hospital, one of Canada’s largest lipid clinics. At the Clinic they conduct next-generation sequencing for lipid metabolism disorders.

Before implementing VS Clinical software in the Clinic, patient data was being gathered without a process in place to deliver the results back to the patients. Since the implementation of VS Clinical, they have been able to generate and deliver patient reports to the clinicians for their annual check-ups with those patients. In addition to the reports, Mark has found the copy number variant (CNV) calling and integrated variant analysis of VS Clinical to be among his favorite features of the software. He has also found the VS Clinical software to simplify the application of ACMG guidelines.

Overall, Mark believes that the Golden Helix software has improved the workflow at the Clinic, referring to it as simple, intuitive, and easy to use. He appreciates having CNV calling, integrated variant analysis, and patient reports contained within one program. Mark estimates that the Clinic was missing approximately 5% of diagnoses prior to using VS Clinical to look at CNV’s.

Mark’s research focuses on the relationship between inflammatory diseases such as sepsis and atherosclerosis and the genetic regulation of cholesterol. He was this year’s first place winner of Golden Helix’s abstract competition. To learn more about Mark’s work, which was recently published in the Journal of the American College of Cardiology, you can read his abstract and view his webinar presentation here.

To request a free trial of VS Clinical email us at info@goldenhelix.com!