Integrating Somatic, Germline and CNV Workflows

with Golden Helix Software

Dr. Bernd Auber is the team leader for the molecular genetic diagnosis of hereditary diseases at the Institute for Human Genetics at Hannover Medical School (MHH). MHH is one of the largest hospitals in northern Germany, with one of the largest outpatient clinics for individuals and families dealing with hereditary cancer and predisposition syndromes.

Dr. Bernd Auber, Hannover Medical School

Dr. Auber became fascinated with medicine in high school after reading the book The Man Who Mistook his Wife for a Hat by the famed neurologist, Oliver Sacks. Having finished medical school, he was trained in neurology and internal medicine and became more interested in research and genetics, which he studied at Gottingen University Hospital. Since 2012, Dr. Auber has been working at MHH, with his most current role focusing on clinical diagnostics for the hospital's patients. There is also a focus on research within his team. Not only are they part of the German Breast and Ovarian Cancer Research consortia, but Dr. Auber's team is also working on a large hearing loss project, in which researchers are working to pinpoint the genetic basis for hearing loss.

The combination of research and clinical focus keeps the human genetics team very busy. Moving forward, Dr. Auber and his team are shifting their focus from exome to whole-genome sequencing, and they hope to be up and running with genome analysis in May 2021. Efficiency is crucial for Dr. Auber, as he wants to decrease the time it takes for NICU patient diagnostics.

NGS data analysis is a complicated process that was time-consuming for Dr. Auber's team. They already surpassed their ability to do the analysis manually and knew they needed to find a better way. Their search for analytical software was narrowed down to tools that could support both germline and somatic data, and this is where Golden Helix's solutions stood out to Dr. Auber and his team. Dr. Auber states, "Golden Helix had little to no competition, as finding other tools that could integrate the workflows for somatic, germline, and CNVs was largely absent from competitor's stacks." Also, a significant selling point to Dr. Auber was the flexibility of the tools. He asserts, "the ability to create unique approaches on how to analyze data that you can individualize in every imaginable way is unique. You are not restricted in any way and can adapt your pipeline to your specific needs". Dr. Auber regards Golden Helix's VSClinical ACMG auto-classifier for variants to be superior to anything else on the market and that the CNV classifier is "something that no other competitor has." For these reasons, MHH decided to utilize Golden Helix.

Dr. Auber hopes to broaden the scope of MHH's diagnostics for both the research and clinical spaces and looks forward to the capabilities provided by Golden Helix that will allow him to re-visit previously analyzed variants in an easy and automated fashion. Dr. Auber and his team are also eager to re-evaluate some of their unsolved cases and to have the confidence that no medically relevant variants will be missed with their new solution.

We look forward to following Dr. Auber and his team at MHH and supporting them in reaching their goals.