Efficient, Scalable and Accessible Solution for

NGS Gene Testing and Variant Discovery

MVZ Martinsried, a German-based leading diagnostics company specialised in hereditary diseases including inherited cancers as well as non-hereditary solid-tumors, implemented Golden Helix's VarSeq Clinical suite to make NGS gene testing and variant discovery efficient, scalable and accessible by converting NGS data output into customized clinical reports in a timely manner.

Since January 2019, MVZ Martinsried is part of Medicover, a leading international Healthcare and Diagnostic Services company founded in 1995. Medicover provides a broad spectrum of healthcare services via an extensive network of ambulatory clinics, hospitals, specialty-care facilities and laboratories through two divisions – Healthcare Services and Diagnostic Services.

Part of the Diagnostic Services, Medicover Genetics is a strategic business unit integrating genetics related businesses from over ten European countries across the division. Medicover Genetics was developed as a greenfield approach, including the acquisition of MVZ Martinsried, to build a strong foundation to become a market leader in genetics. Operating now on a global scale, while expanding the samples analysis volume, MVZ Martinsried sparked the need for a more robust diagnostics pipeline and a solution that could accommodate different locations with local installation.

Dr. med. Konstanze Hörtnagel, Deputy Head of Human Genetics at MVZ Martinsried, spearheaded the project and began to evaluate commercial options supporting Next-Generation Sequencing (NGS) data. "Our previous interface was clunky and inefficient for the analysis volume our lab was starting to receive. VarSeq stood out during our initial round of evaluations with all its tools being integrated in one workflow," Hörtnagel notes.

Her team evaluated Golden Helix's VarSeq Clinical Suite enjoying the amount of flexibility the platform offered their lab through implementing their own preferences, filtering and annotation, and the ease of transition from sequencing data to report. Adding Copy Number Variant (CNV) calls directly from NGS data into the workflow side-by-side with Single Nucleotide Variants (SNVs) was also a plus. VSClinical's consistency across users and automation made it easy for her team to share information across MVZ Martinsried's lab and other locations within Medicover Genetics. Additionally, VSClinical's integration of both germline and somatic workflows streamlined their clinical pipelines. The lab output is now stored in VSWarehouse for databasing where they were able to build their own variant allele frequency statistics enabling any new variant to be annotated and potentially filtered with the frequency of that variant in their VSWarehouse projects.

One of the most crucial factors that would contribute to her decision was a solution's ability to scale with her lab's growing volume with abiding support; Hörtnagel needed to ensure she chose a solution that would serve her lab for the long haul. "Our decision to implement Golden Helix's Clinical Suite came down to the support we received throughout the evaluation. During the implementation phase and beyond continued solution-orientated assistance ensures our laboratory requirements are met," she elucidates. "Golden Helix does that for you so your team can spend their time and focus on getting results to the patients."