Transforming Precision Medicine in India

with Golden Helix Software

Established in 2004 and headquartered in Chennai, India, with regional centers across the country, LifeCell runs India's largest stem cell bank and has also diversified into diagnostics and tissue therapeutics. They employ roughly 2,000 people, providing genetic services to customers in the mother and baby space.

Phani Nagaraja Setty is working as scientist at LifeCell. Setty obtained his Master's degree in biochemistry and then received his Ph.D. in genomics with a concentration in Type 2 diabetes. For the last three and a half years, he has managed the lab operations for NGS tests in Lifecell diagnostics. While working on NGS based clinical exome panels, he realized the need for an efficient genotype-phenotype correlation database orienting to the Indian population. LifeCell committed to a goal: to democratize genetic testing across the country thus making an efficient translation of genomic information in clinical decision.

Setty and his team offer tests that are some of the first of their kind in India. They have developed NICU babies' panels, infertility, and testing to follow up their prenatal reflex DNA testing. Any newborn babies that come back positive from a biochemical screening of over 20 common inborn errors of metabolic disorders get a free genetic reflex test.

When Setty began shopping around for commercial software solutions to handle this testing volume, he was able to leverage his prior experience working in software development. He knew specifically what he wanted in terms of the genomic workbench. After evaluating five different products, he decided that Golden Helix's clinical variant interpretation platform contained all the key attributes that he knew were essential for success. He appreciates the scalability and usability of the tools and especially finds value in being able to analyze SNV and CNVs in the same workflow. Setty cites VS-CNV as "the most refined CNV calling solution that I have seen."

It also helped LifeCell maintain its commitment to the people of India. By using Golden Helix, they could eliminate the additional expense of MLPA testing for the confirmation of CNVs in most cases where direct MLPA testing is not possible. Setty explains that identifying CNVs in x-linked disorders is very difficult and said that 90% of their Congenital Adrenal Hyperplasia (CAH) and Duchenne Muscular Dystrophy (DMD) cases were resolved by using Golden Helix's approach of merging SNV and CNV analysis. Setty's testament, "The performance of Golden Helix's CNV algorithms was a bullseye for the identification of the CNV's in the X chromosome. The results were completely matching the earlier MLPA results for certain genetic conditions, and Golden Helix was absolutely the right solution for LifeCell," speaks for itself.

Other features of the software that LifeCell's team greatly appreciated were the tools' scalability and usability. Golden Helix's customer support is also very appreciated by LifeCell. Setty's previous requests for a clinical reporting Word-based format were heard and delivered.

LifeCell has emerged as a leader in diagnostic testing, stem cell banking, and tissue therapeutics in a very short time. Their pre and post-test counseling services are unparalleled. This innovation and commitment to making genetic testing an affordable and accessible tool for the Indian population has created tremendous growth for their company. LifeCell's goal for 2021 is dedicated to whole-genome sequencing. Setty plans to meet this goal by leveraging the full stack of tools from Golden Helix. Their future road map includes female cancer panels, childhood cancer panels, somatic mutations, and transcriptome-based analysis.