In the course of designing more than 200,000 primer pairs and generating more than 18 million sequence reads, bioinformatics specialists and R&D scientists who worked on the Appliera Genome Initiative gained valuable experience in primer design, as well as insight into the genomic complexities and technical challenges associated with them. The 3130 and 3130xl Genetic Analyzers, when combined with the Applied Biosystems VariantSEQ™ Resequencing System and SeqScape® Software v2.5 for seamless data analysis, provide the most robust and efficient resequencing system for customers who require low to medium throughput. The components of this integrated system simplify the resequencing steps found in current resequencing protocols, and offer a complete, cost-effective solution for laboratories performing either large or small resequencing studies, thus, revolutionizing DNA sequencing as a tool for the detection of human gene mutations.

A HIGH PERFORMANCE SYSTEM FOR SEQUENCING APPLICATIONS

The 3130 Series Genetic Analyzers are fully automated, high-performance, fluorescence-based, capillary electrophoresis systems that can analyze multiple samples simultaneously. Samples can be resequenced in 35 minutes when using the 36-cm capillary system or in 44 minutes using the 24-cm capillary array. After the sequence data is collected, the KB® Basecaller will automatically process the data to produce a length of read (LOR) greater than 500 base pairs (bp). Furthermore, the throughput of the 3130 system, using the UltraILix® POP-7™ run module, can efficiently sequence up to 41 runs (696 samples) in a 24-hour period, generating high-quality results with minimal hands-on time.

The 3130 series system is designed for ease-of-use to maximize laboratory productivity while reducing the overall cost per sample. Now more than ever, researchers have the flexibility to choose one configuration for all their resequencing needs. The Automated Polymer Delivery System in the 3130 series system allows automatic polymer loading, which minimizes hands-on time and maintenance while maximizing performance. The system enables the use of 3130 POP-7 polymer, not only for the 36-cm capillary array, but also for the 50cm and 80-cm capillary arrays. The run configurations, specific for 3130 POP-7 polymer, incorporate a higher temperature through the detection cell heater, which yields less run-to-run variability, and faster electrophoresis times than any other capillary electrophoresis system available on the market today.

KEY FEATURES OF THE 3130 SERIES SYSTEMS

- Automated Polymer Dispenser
- New 3130 POP-7™ Polymer
- One polymer & one array for any application
- Faster turn around times
- Longer Reads
- Better peak resolution
- Easy to use
- Easy to maintain
- Automated Analysis using SeqScape® Software v2.5
- Dynamic report generation
- VariantSEQr™ Resequencing System: The PCR process is covered by patents owned by Roche Molecular Systems.

The process is completely automated providing fast and accurate data analysis. After the primers have generated sequences, the sequence files and project template (part of the resequencing primer product) are analyzed by SeqScape® software (Figure 2).

Applied Biosystems provide project templates for each Resequencing Set (RS). Each project template provides a reference sequence (Reference Data Group) to which all the specimens in a project are compared, settings that are used to analyze the data (Analysis Defaults) and settings that are used to display the data (Display Settings). By directly importing the RS project template into the SeqScape Manager within the software, a project can be created. After the project is analyzed, the quality of the results can be reviewed and the variants can be examined (Figure 3).

**RESULTS**

The 3130 series systems provides seamless integration between the instrument and analysis software, ensuring automatic sample loading, generation of sequencing data, basecalling, and alignment of sequence and reference data. An integral part of successful resequencing, SeqScape® Software analyzes data from both small- and large-scale projects.

**CONCLUSION**

To address the challenges inherent in the use of DNA sequencing for mutation detection, Applied Biosystems developed an integrated system that comprises the new 3130 series Genetic Analyzers, the VariantSEQ™ Resequencing System, and SeqScape® software. Together, this system, along with the BigDye® Terminator chemistry kits, provides a complete, cost-effective solution for mutation detection.