



GENEREADER

CASE STUDY

THE CHALLENGE

The capacity to sequence all 3.2 billion bases of the human genome has increased from 1.3 human genomes sequenced annually to 18,000 human genomes a year within a decade.. Despite this improvement sequencing a genome is still an extremely complex procedure, which prevents its broad use in routine human diagnostics.



THE GOAL

Build a complete NGS workflow with seamlessly integrated automated components offering ease of use and efficiency from sample to result. Provide actionable insights with validated gene panels and fully integrated bioinformatics.



THE RESULT

The Genereader is the only integrated NGS workflow from sample to insight..



THE APPROACH

The GeneReader workflow includes the following 6 processes: sequencing primer hybridization, flow cell preparation, reagents preparation, experiment set-up, flow cell loading and run start, and post-run maintenance wash. The sequencing chemistry consists of terminator-dNTP sequencing-by-synthesis. After the DNA library construction, DNA is clonally amplified using the GeneRead QIAcube and immobilized via direct bead-slide interaction and exposed to a DNA sequencing primer to produce a high-density array on a Flow Cell. To read out the sequence of each of the beads, the array of fragments is first subjected to reagents containing DNA bases that include a removable fluorescent dye and an end cap. These bases attach themselves to the end of the growing strand of DNA in accordance with the base on the complementary strand. The array is scanned by a high-resolution electronic camera and the fluorescent output of each of the four dye colors at each array position is measured and recorded. The color indicates which base (A, C, G or T) was incorporated onto the DNA fragment from the previous step. Finally, the array is exposed to cleavage chemistry to break off the fluorescent dye and end cap that will then allow additional bases to be added. This cycle is then repeated on the GeneReader.