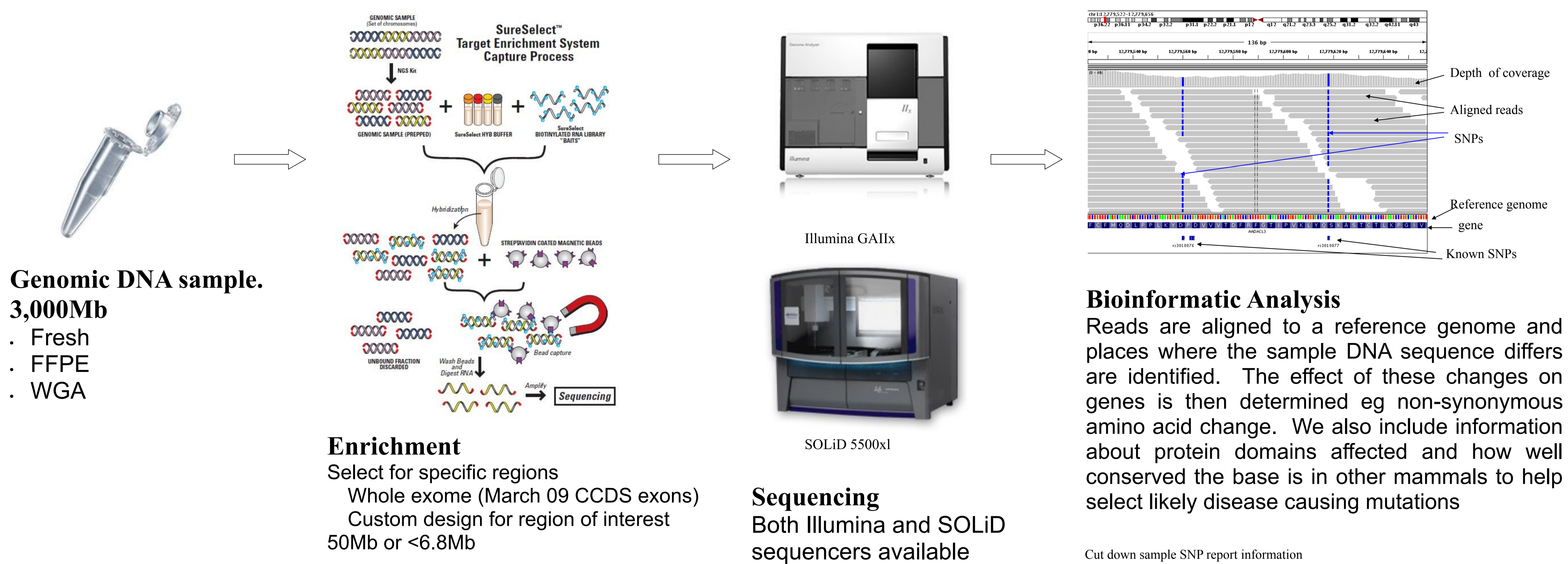


Next Generation Sequencing in Clinical Genetics with the EASIH

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Next Generation sequencing has revolutionised many areas of molecular biology and genetics research. Technological advances have brought the cost of DNA sequencing down to sufficient levels that sequencing the coding regions of human genomes in many patients is now feasible. This “whole exome” approach to disease mutation identification has been successfully demonstrated in many cases [1,2,3]. The Eastern Sequencing and Informatics Hub (EASIH) based at the University of Cambridge is a not-for-profit research facility available for researchers throughout the country to take advantage of Next-Gen sequencing without having to invest large sums in infrastructure and expertise. After set up costs, one of the other major hurdles associated with Next Gen sequencing is the data analysis but we have a team of bioinformaticians available to work on each project. Our expert bioinformatic analysis can filter out many thousands of variations so that you can focus effort on strong candidate genes.

Whole exome / custom enrichment and sequencing



Smaller but Faster Sequencing



Other applications of Next-Gen sequencing include amplicon sequencing of large cohorts, chromosomal structural alterations and sequencing of custom sets of genes. We have recently acquired an Ion Torrent Personal Genome Machine which has a very rapid turnaround time and so is potentially very interesting in the clinical context. It's current 20Mb capacity is well suited to amplicon sequencing and bacterial genomes but we expect this rapidly rise over the next year. The Roche GS Junior is also targeted for clinical applications



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1) Miller Syndrome - Nat Genet. 2010 January; 42(1): 30-35.
2) Bartter Syndrome - Proc Natl Acad Sci U S A. 2009 106(45): 19096-19101.
3) Charcot-Marie-Tooth - Ann Neurol. 2011