# FREE OUR GENES: A LIBERAL RELIEF

## DID YOU KNOW THAT IT IS POSSIBLE TO SEQUENCE YOUR HUMAN GENOME FOR $1,000?

HiSeq X Ten, a new $10 million gene-sequencing machine developed by Illumina, can carry out more than 2 genome sequences an hour, each one sequenced 30 times for accuracy. If one were to compare, mapping the first human genome took over a decade and cost $3 billion. Illumina explained that the cheap price of $1,000 is caused by the projected volume, i.e. 18,000 sequences/year. The price consists of $797 for the reagents used during the process, machine depreciation of $137 and employee overheads of $65.

Although the price at which the sequencing is effectively being marketed depends on high volumes, it seems that the price tag is not factoring in the mark-up, costs of analyzing the completed genome or storage costs. These are extremely very expensive add-ons. Nonetheless, even if the actual price will be higher, we seem to be drawing nearer to start sequencing human genomes in numbers large enough which will allow a direct comparison between them, leading to a translation into personalized medicines. The raison d'etre is that most diseases are the result of many genes, each contributing a small amount to disease development. Actually, by 2017, the UK plans to sequence more than 100,000 patients within its National Health Service. This project was heralded in June 2013 through the formation of *Genomics England,* which spearheads this £100 million project.

The importance of this technology is aptly elucidated by the following scenario. Today, Shelby Valint is a 14 year old Phoenix girl, largely leading a normal life. However, 5 years ago she was bound to a wheelchair, having struggled against an unknown debilitating illness for 9 years. It was only after researchers mapped her entire genome that a defect was identified in the Dopa Decarboxylase gene. This defect prevented the production of sufficient amounts of dopamine. She was eventually successfully prescribed bromocriptine and selegiline, eventually substituting bromocriptine with pramipexole.

Apart from these exceptional cases, one of the possible routine uses of this technology is in pregnancy since sequencing can help identify fetal abnormalities in maternal blood samples. A further application is in tumor mapping. Interestingly, the handheld molecular diagnostics device pioneer QuantuMDx Group has secured a $8.42m funding which will be utilised to optimize and trial Q-POC™, a handheld DNA sequencing & genotyping device. Basically, this $1,000 device can analyse the DNA from a drop of blood/sputum in 15 minutes. This year, field trials will be conducted to genotype the *Plasmodium falciparum, P. vivax, P. ovale, P. malariae and P. knowlesi* with a view to implement drug-resistance strategies. Also this year, Q-POC™ will be used within the UK's National Health Service in order to identify any genetic variations which may affect their response to warfarin.