Presentation title: The future of DNA sequencing technology

Abstract:
I will review the recent history of “Post-Sanger” sequencing technology, and then make some wild and unjustified extrapolations into the future based on too few data points.

I will review some of the technologies on the horizon and ask how we can appraise them.

For example, if we can define sequence read quality as a composite of read length and base-calling accuracy, recent trends have overwhelmingly been in the direction of quantity at the expense of quality. As a consequence a great deal of informatics effort has been expended in managing rather poor quality data. Of course the human genome, along with many other genomes, is not particularly amenable to analysis, contain entities such as pseudogenes, non-coding regions (sometimes referred to as “junk”, sometimes claimed to be functionally important) and short repeats. So how does the collision of a relatively refractory analyte like the human genome and an imperfect sequencing method result in a “genomics revolution”? What have we gained and what are the current limitations that need to be addressed in future technologies?

I will look at two examples of the impact of current and pending sequencing technology: tumour analysis in fixed and fresh tissue and the identification of allele expansions.