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Editorial

Next Generation DNA Sequencing Technologies

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Next generation sequencing (NGS), massively parallel or deep sequencing are related terms that describe a DNA sequencing technology which has revolutionised genomic research. In contrast, the previous Sanger sequencing technology, used to decipher the human genome, required over a decade to deliver the final draft. Although in genome research NGS has mostly superseded conventional Sanger sequencing, it has not yet translated into routine clinical practice. The aim of this article is to review the potential applications of NGS in paediatrics.

There are a number of different NGS platforms using different sequencing technologies, a detailed The main utility of NGS in microbiology is to replace conventional characterisation of pathogens by morphology, staining properties and metabolic criteria with a genomic definition of pathogens. The genomes of pathogens define what they are, may harbour information about drug sensitivity and inform the relationship of different pathogens with each other which can be used to trace sources of infection outbreaks. The last recently received media attention, when NGS was used to reveal and trace an outbreak of methicillin-resistant Staphylococcus aureus (MRSA) on a neonatal intensive care unit in the UK.4 What was most discussion of which is beyond the scope of this article. However, all NGS platforms perform sequencing of millions of small fragments of DNA in parallel. Bioinformatics analyses are used to piece together these fragments by mapping the individual reads to the human reference genome. Each of the three billion bases in the human genome is sequenced multiple times, providing high depth to deliver accurate data and an insight into unexpected DNA variation (figure 1). NGS can be used to sequence entire genomes or constrained to specific areas of interest, including all 22 000 coding genes (a whole exome) or small numbers of individual genes.

Remarkable was that routine microbiological surveillance did not show that the cases of MRSA that occurred over several months were related. NGS of the pathogens, however, allowed precise characterisation of the MRSA isolates and revealed a protracted outbreak of MRSA which could be traced to a single member of staff.

The main disadvantage of NGS in the clinical setting is putting in place the required infrastructure, such as computer capacity and storage, and also the personnel expertise required to comprehensively analyse and interpret the subsequent data.

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