British Thoracic Society Genomics and Repsiratory Medicne

Why the NHS has embraced Genomics?

Medicine is moving from disease-based approaches, where trials of thousands of individuals are used to identify best practice, to a personalised, tailored approach for each patient. Personalised medicine will provide patients with better opportunities for diagnosis and treatment, reducing diagnostic delays and avoiding unnessecary and potentially harmful investifations or treatment side effects.

What are the opportunities?

Genomics offers the clearest route to date to tailor pathways to best suit the individual. Genomics refers to the analysis of DNA - not just the 20,000 or so genes, but all of the other regulatory and even non coding regions with DNA ("the genome"). This may be the DNA from an individual that they inherited from their parents, DNA that has undergone changes in developing into a cancer for example, or the DNA of a microorgansim causing important infection. Such DNA influences and sometimes determines precise pathophysiology. Increasingly, identifying minor differences in the DNA sequence allows us to identify which exact pathology is present, and why; which treatments might have greatest impact; and which drugs are more likely to cause harm for that particular individual. Sequencing the whole genome provides the required blueprint at a realistic cost, and is predicted not only to offer substantial benefits to patients, but also, to overall cost-saving for the NHS.

Relevance to Respiratory Medicine

Many respiratory patients are potentially eligible to have Whole Genome Sequencing (through 100,000 Genomes Project) as a prelude to improved personalised care in the future. This is available to those without a known molecular diagnosis‡, in key groups:

1. Bronchiectasis (if more than one case in immediate family*, multilobe disease, and not attributable to other infective or respiratory pathology)
2. Primary ciliary dyskinesia
3. Idiopathic pulmonary fibrosis (if more than one case in immediate family*, no cystic disease, and no other respiratory disease present)
4. Familial Primary Spontaneous Pneumothorax (if more than one case in immediate family)
5. Pulmonary arteriovenous malformations (if multiple, more than one case in family, and/or underlying hereditary haemorrhagic telangiectasia (HHT)
6. Severe multi-system atopic disease (if high IgE, >5,000 IU/mL)
7. Sleep apnoea patients (if inherited sleep disorders or syndromic causes of obesity)
8. Patients with chronic respiratory failure (if due to neuromuscular disease or thoracic dystrophies)
9. Immunodeficiency (sepcified B cell, T cell, granulocyte, neutrophil and/or complement deficiencies)

‡: the causative gene should not be already known in the family
* First degree relative, i.e. parent, sibling or child