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From cardiovascular genomics to interdisciplinary precision medicine

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Human primary or essential hypertension is a complex, polygenic trait with some 50% contribution from genes and environment. Richard Lifton and colleagues provided elegant dissection of several rare Mendelian forms of hypertension, exemplified by the glucocorticoid remediable aldosteronism and Liddle's syndrome. These discoveries illustrate that a single gene mutation can explain the entire pathogenesis of severe, early onset hypertension as well as dictating the best treatment.

The dissection of the much more common polygenic hypertension has proven much more difficult. The real breakthrough came with the initiation of the genome wide association studies (GWAS) characterised by a much more thorough coverage of the genome with thousands single nucleotide polymorphisms (SNPs). Typically 500,000 – 2,5000,000 SNPs have been used for the big, collaborative GWAS for hypertension. These studies resulted in several "hits" or signals with a genome-wide significance and a high level of reproducibility between studies. These "hits" have been used successfully to calculate genetic risk scores for cardiovascular complications such as left ventricular hypertrophy, stroke and coronary artery disease. Intragenic signals, such as for example Uromodulin, are being used to examine new pathways for cardiovascular protection and possibly new targets for drug discovery as well as new style stratified clinical trials.

The next steps in genomic medicine belong to a combination of the next generation sequencing (NGS) and/or other "omics" data followed by linkage with electronic health records, including preferably the real time clinical data, biochemistry, imaging, histology as well as longitudinal health outcomes.

Precision medicine involves examining the genetic makeup of patients and their differing responses to drugs designed to treat specific diseases. By building up an understanding of the 'strata' of responses and the genetics of the diseases, we hope to create more personalised and effective forms of treatment for groups of patients most likely to benefit. Significant past investment in Scotland in electronic health records (EHRs) and translational medicine research, coupled with a vibrant healthcare technology industry, positions Scotland as the location to drive forward the precision medicine agenda globally.