Twenty-first century genomics for sports medicine: what does it all mean?

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We can easily be left behind by the explosion in technological developments in medicine—or is it just a sign of getting older? Many of us will remember a time before MRI were readily available, and having to learn what fat suppressed images, T1s and T2s actually meant.

Genomics is another example of a rapidly expanding technological development with major implications for medicine, and even experienced clinicians (many without formal training in molecular biology) are struggling to understand this. It requires learning a whole new language. The sequencing of the 3.2 billion nucleotides that compose the human genome was first completed only in 2003, at an estimated cost of $2.7 billion. At the time, it was simply something of interest, but Bouchard shares a clear alternative contention that a complex system can be more easily understood if considered as a whole. Claude Bouchard reflects on the complexity of exercise genomics and how little we really know. The research to date has failed to adequately deliver results, but Bouchard shares a clear alternative view for future research.

**GENOMICS LITERACY—WHAT DOES IT ALL MEAN?**

Genomics is finding its place in medicine, from genetic testing for rare disorders to personalised medicine based on our genetic make up (see page 1517). However, the role of most of the genes in the human genome is still unknown.

A ‘snip’ is no longer just a glib term for vasectomy, because in genomics, an SNP (pronounced ‘snip’) is a ‘single nucleotide polymorphism’. SNPs are locations within the human genome where the type of nucleotide present can differ between individuals. You also need to know Genome-wide Association Studies (GWAS) from candidate gene study in the new world of ‘Omics’. Candidate gene studies test the validity of an ‘educated guess’ of one or more variants within the candidate gene in relation to a particular condition, for example, Achilles tendinopathy and the COL1A1 gene, which encodes for the α1(I) chain of type I collagen (see page 1497). They were predominantly used before testing of the whole genome became easily available. GWAS examine SNPs across the genome, looking for associations with particular conditions. Scanning the whole genome is a broad fishing expedition approach without any prior assumptions.

‘Omics’ technologies (figure 1) are aimed at the universal detection of genes (genomics), messenger RNA (mRNA) (transcriptomics), proteins (proteomics) and metabolites (metabolomics), with the contention that a complex system can be more easily understood if considered as a whole.
that these companies quote), is that, at present, DNA testing has no role. SEM
physicians need to discourage any parent, child or coach from using these tests (see
page 1486).

FROM THE LABORATORY TO THE
CLINIC—WHAT ROLE FOR
GENOMICS?
What is the role of ‘Omics’ in sport? How
do we keep up with the essentials? We are
practising in the age of personalised SEM;
personal whole genome sequencing, per-
sonal health records and personal technol-
ogical health applications (smart phones,
etc). The world is getting smaller. This is
good for partnerships forcing us to com-
municate and collaborate—with each
other, with the research community and
with patients.

There clearly needs to be a willingness to
collaborate—to advance not only the
science but also the translation to concrete
guidelines for clinical SEM practice. New
initiatives are required to address these de-
fi ciencies in our knowledge, and to develop
practical and valid guidance for SEM.
Professor Claude Bouchard highlights the
danger of performing sports genetics
research with limited subject numbers (see
page 1492).

Large collaborative studies with appropri-
ately phenotyped subjects, incorporating
genome-wide approaches may be able to
deliver some answers. In time, we may
understand how to use this information to
safely guide athletes. What is clear is that
SEM practitioners in the 21st century need
to engage in the genetics of sport, exercise
and injury—now.

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Definitions

Genome: The total DNA of a cell or organism
Polymorphism: Variations in DNA at a specific site
Transcriptomics: The study of the mRNA within a cell or organism
Transcriptome: The total mRNA in a cell or organism
Proteomics: The large-scale study of proteins, including their structure and
function, within a cell/system/organism.
Proteme: The set of all expressed proteins in a cell, tissue or organism
Metabolomics: The study of global metabolite profiles in a system (cell, tissue
or organism) under a given set of conditions
Metabolome: The total quantitative collection of low-molecular-weight
compounds (metabolites) present in a cell or organism that participates in metabolic
reactions.

Genome Wide Association Studies: Their purpose is to determine alleles that correlate to
different diseases and traits with a hypothesis free approach.

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