WHY IS THE PREVALENCE OF KNEE OSTEOARTHRITIS INCREASING?

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Invited speaker abstract submission
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D. Felson
I will give my lecture on: Friday, 15 June 2018
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PHYSICAL ACTIVITY AND EXERCISE: OPPORTUNITIES AND CHALLENGES

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Osteoarthritis (OA) is a steadily growing public health concern in particular as there is still a lack of curative therapeutic options in a biological sense. Thus, prevention of OA becomes an increasingly important topic. Physical activity, and exercise are among the key modifiable risk factors associated with OA. Joins are built to be used, but overuse and joint injuries are also linked with incident OA. This presentation will provide some key opportunities with exercise to maintain joint health, but also the challenges when the window of opportunity is exceeded.

Disclosure of Interest: None declared

BIG DATA FOR TREATMENT EFFECTIVENESS AND SAFETY

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While randomised clinical trials remain the gold-standard for examining drug-effects, there are limitations to the evidence they provide. For example, studies are usually too small or too short to detect rare and long-term adverse effects. This is particularly true for kidney-related outcomes where many trials were underpowered to detect outcomes of interest such as end-stage renal disease, too historic to collect data on newly-defined entities such as acute kidney injury or simply excluded patients with chronic kidney disease. In addition, disproportionate recruitment of Caucasian men in mid- to older-life limits the evidence base for understanding risk of adverse-effects in other population groups such as women, the elderly and people of non-Caucasian ethnicity. Determining these outcomes requires long-term surveillance of population databases, usually anonymised health care records, once a drug is in routine use.

In this talk I will discuss recent work from our group where we have used routinely collected renal function measurements from primary care to precisely examine kidney-related and other adverse effects. I will discuss some of the difficulties of this approach as well as the potential it offers to understand better the balance between risks and benefits of many drugs in common use.

Disclosure of Interest: None declared

FROM MENDELIOME TO PERSONALISED MEDICINE IN CHILDLHOOD SLE

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Pediatric-onset SLE is a rare condition where genetics traits are believed to play an important causal role. A few monogenic causes of SLE have been described mostly in familial forms, including complement deficiencies, so-called type I interferonopathies, and B cell related defects. Genetic studies in mouse models and genome-wide associations studies in patients have also pointed to other genes potentially involved in juvenile SLE. However, large-scale sequencing analyses of paediatric SLE are lacking, and the overall contribution of genetic factors in disease onset is therefore unknown. Personalised medicine relies on the understanding of underlying mechanisms. Genetic discovery and functional characterization of the variants prefigure the precision medicine in complex diseases such as SLE.

We have designed a NGS panel comprising genes for which mutations are known lupus causing (KLC) also reported as the Mendelio) as well as prospective candidate genes, potentially lupus causing (PLC), and analysed 117 children who fulfilled ACR criteria for SLE from two large cohorts of pediatric-onset SLE in the UK and France. Genetic variants were identified and filtered to select rare (ExAC database frequency of <1% for homozygous variants and <0.01% for heterozygous variants) and predicted in silico as damaging by different algorithms.

We identified mutations in KLC genes for 8 patients. Variant segregation within families and functional analyses supported the causal role of these mutations. Other patients had monoallelic variants in recessive KLC genes, which may have contributed to disease onset and other patients displayed rare and pathogenic variants in PLC genes. This enrichment was specific to the lupus cohort compared to a control cohort of healthy patients.

Against this backdrop of growing data and increasing computational resources, the use of data science methods including machine learning is becoming popular for analysing large-scale medical datasets. This talk provides a brief overview of machine learning methods for healthcare applications including an introduction to supervised and unsupervised learning, followed by real-world examples of data analysis using machine learning, such as (a) the development of prognostic models for clinical risk assessment, and (b) mining of electronic health records for detecting patterns and phenotypes within a population.

Disclosure of Interest: None declared

HOW BIG DATA AND MACHINE LEARNING COULD CHANGE THE GAME

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With recent advances in the acquisition and digitisation of medical data, the use of routinely collected health data for research is on the rise. Routinely collected data comes with the promise of the 3 V’s of “big data”: volume, velocity, and variety. Recent recommendations by the UK National Institute for Health and Care Excellence and the Academy of Medical Sciences have therefore acknowledged the potential for data science methods to play an increasingly important role in healthcare research.

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From big data to personalised medicine in paediatric rheumatic diseases

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