Why is the prevalence of knee osteoarthritis increasing?

D. Felson. Clinical Epidemiology Unit, Boston University School of Medicine, Boston, USA

Invited speaker abstract submission

EULAR18-7734

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I will give my lecture on: Friday, 15 June 2018

Disclosures of Interest: None declared


Prevention of OA: yes we can!

FRIDAY, 15 JUNE 2018

SP0123

WHY IS THE PREVALENCE OF KNEE OSTEOARTHRITIS INCREASING?

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Big data for musculoskeletal research

FRIDAY, 15 JUNE 2018

SP0125

HOW BIG DATA AND MACHINE LEARNING COULD CHANGE THE GAME


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 health care research.

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

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From big data to personalised medicine in paediatric rheumatic diseases

FRIDAY, 15 JUNE 2018

SP0127

FROM MENDELIOME TO PERSONALISED MEDICINE IN CHILDHOOD 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 Mendelome) 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.