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

Disclosures of Interest: None declared


PHYSICAL ACTIVITY AND EXERCISE: OPPORTUNITIES AND CHALLENGES

M. Englund, Clinical Epidemiology Unit, Orthopaedics, Department of Clinical Sciences Lund, Lund University, Lund, Sweden

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. Joints 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.

Disclosures of Interest: None declared


FROM BIG DATA TO PERSONALISED MEDICINE IN PAEDIATRIC RHEUMATIC DISEASES

A. Belot1,2, on behalf of Genial/Lumogene working group. 1Pediatric Rheumatology Unit – National Referee Centre for Rheumatism and Autoimmune diseases – Raise, Chu Lyon; 2U111, INSERM, Lyon, France

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 understanding underlying mechanisms. Genetic discovery and functional characterisation 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 Mendelrome) 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.

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FROM MENDELOMIE TO PERSONALISED MEDICINE IN CHILDHOOD SLE

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