GBSBC

Genetics, Bioinformatics, & Systems Biology Colloquium

presents

Andrew Jackson, MBBCH

Professor

MRC Human Genetics Unit, Edinburgh

JOIN US ON ZOOM!

THURSDAY
JUNE 1

12PM

LEICHTAG AUDITORIUM

ZOOM

JOIN

Mendelian genetics in a Post-Genomic age: AGS to cancer genomics.

Sydney Brenner's 2002 Nobel prize lecture envisaged humans as the model organism of choice in an age of genomics. Since, comprehensive discoveries have been made for single gene disorders, but have been outpaced by the delineation of millions of risk variants for common complex diseases, and extensive mutation catalogues of cancers. As a clinical geneticist, my expertise is monogenic disorders. Through my work on such conditions, I have developed a fascination for how the study of ourselves can provide insights to fundamental biological processes and disease pathogenesis. This seminar will outline our work on a rare neuroinflammatory disorder of childhood, how it led us to discover the most common nucleotide lesions in the mammalian genome, link genome instability to innate immune activation, and to investigate the mechanistic basis of cancer mutation signatures. Through this, I hope to persuade you that rare genetic disorders have continuing utility in our ambitions to understanding human pathophysiology, complementing genomic approaches in cancer and common disease.

PRESENTED BY:

IGM Institute for Genomic Medicine

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