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Genetics in research – towards understanding pathophysiology

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Large-scale genome-wide association studies have now identified hundreds of genetic variants of interest that relate to kidney function and damage, but the mechanisms by which the variants affect the kidney remain uncertain. The integration of multi-modal omics data can help elucidate the underlying pathophysiology. In this lecture, we will discuss several examples of integrative omics as they relate to kidney disease. First, we illustrate the evaluation the downstream, potentially mediating effects of genetic variants. Second, we discuss integration of genetic effects as instrumental variables via techniques such as Mendelian Randomization to build support for causality of biomarker-phenotype associations. Third, we discuss the combination of omic layers to help with annotation and target gene prioritization.