Contribution ID: **12** Type: **not specified**

Keynote 5, Prof Liz Carpenter: Using Structural biology of human membrane proteins to understand the causes of genetic diseases

Thursday 10 October 2019 10:00 (40 minutes)

The human genome project and subsequent sequencing efforts of thousands of patients and healthy individual provides a wealth of associations between variants in genes and diseases. At the SGC our aim is to create a step-change in drug development by providing tools (proteins, structures, assays and bound small molecules), for proteins that are have associations with genetic disease. We focus in particular on proteins that are associated with neuropsychiatry, cancer, rare and metabolic disease, as well as inflammatory conditions. These "Target Enabling Packages"or TEPs, are made freely available to advance our understanding of the biology of disease and to assist in the design of therapeutics. The Carpenter group focuses on integral membrane proteins, including ion channels, solute carriers, ABC transporters and enzymes. Here, I will discuss three examples of genetic hits, PKD2 in kidney disease, TMEM16K in ataxia and DPAGT1 in congenital myasthenia, for which we have obtained structures, and a wealth of additional improvement in our understanding disease biology. These examples of structures of genetic hits illustrate the power of structural biology, as well as the need for extensive additional information, to provide an understanding of disease biology, which is essential for development of therapeutics.

Presenter: Prof. CARPENTER, Liz (University of Oxford, UK)

Session Classification: Keynotes and Short talks 3