Ryan Yuen- Lay Summary

Autism spectrum disorder (ASD) is one of the most common neurodevelopmental disorders with a strong genetic basis. However, the genetic contributing factors identified so far can only explain about 20% of cases, which is lower than the family studies indicated. Using a novel approach to search for the missing genetic contributing factors from the DNA sequence, our group has discovered a set of previously unexplored repetitive sequence that tend to increase in length (i.e. repeat expansions) in ASD. These repeat expansions were found in genes that are responsible for nervous system development, but the mechanism of how they impact the related gene's function is mostly unknown. This study aims to address this and elucidate this gap in knowledge with the use of cellular and animal models. We hypothesize that the repeat expansions may affect RNA processing that leads to abnormal development and function of the central nervous system. By understanding the mechanism of how repeat expansions are involved in ASD, we would like to eventually broaden the scope by which ASD and its related conditions may be treated.