September 2021

The role of prr12a and prr12b in Zebrafish: A Model for a Rare Human Disease

Renee J. Resendes BSc
Western University, rresend@uwo.ca

Gregory M. Kelly PhD
The University of Western Ontario, gkelly@uwo.ca

Tugce Balci MD, FRCPC FCCMG
The University of Western Ontario, tugce.balci@lhsc.on.ca

The role of prr12a and prr12b in Zebrafish: A Model for a Rare Human Disease

Pathogenic variants in the human proline-rich 12 (*PRR12)* gene are believed to cause a neurodevelopmental disorder based on a case series of 24 unrelated individuals. The *PRR12* gene is believed to play a role in neural development due to its enriched expression in the eyes, pituitary gland and cerebellum; however, little is known about the exact function of the *PRR12* gene and protein. In order to elucidate this, studies using model organisms such as zebrafish are needed. The zebrafish is a powerful model system to study genes involved in development and disease owing in part to their fast generation time and amenability to a plethora of molecular genetic techniques. My research entails characterizing the *prr12* genes *(prr12a and prr12b)* in zebrafish; to understand how these genes are regulated during embryonic development and establishing a *prr12a/12b* zebrafish mutant line to corroborate the phenotypic abnormalities seen in patients with the *PRR12*-related disorder.