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

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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 *prp12* genes (*prp12a* and *prp12b*) in zebrafish; to understand how these genes are regulated during embryonic development and establishing a *prp12a/12b* zebrafish mutant line to corroborate the phenotypic abnormalities seen in patients with the *PRR12*-related disorder.