

Lay Summary

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Social Neuropeptide Dysfunction in Fragile X Syndrome

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Fragile X Syndrome (FXS) is a genetic disorder that causes abnormalities in brain development. FXS patients show the following symptoms: social deficits, mental retardation, hyperactivity, impairments in language, and the performance of repetitive behaviours. The disorder is life-long. There is no cure, nor is there any type of intervention that can correct the brain abnormalities that cause FXS. The changes that occur in the brains of FXS patients are very poorly understood. By understanding how the brain is altered in FXS, researchers can develop therapeutics that are designed to repair the brain changes that mediate FXS symptomology and thereby ameliorate symptoms. Social deficits are one of the most characteristic and profound symptoms shown by FXS patients, which is very similar to Autism. Here, I am proposing to study the regions of the brain that regulate social behaviour, and to determine if this region of the brain is abnormal in FXS patients. This has never been investigated directly in FXS or Autism. To do so, I will investigate changes in brain cells that utilize the chemicals oxytocin and vasopressin, as well as brain cells that receive signals from these chemicals, in post-mortem brain tissue from FXS patients.