

## **Lay Summary**

**BRAIN UK Ref: 17/011**

### **Tau and A2AR expression in Alexander's disease**

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Alexander's disease is a rare terminal disease mostly affecting children. The disease is in a group of brain cells called astrocytes and a genetic link has been identified as the cause. However, little is known about how the genetic problem leads to disease.

One hypothesis is that astrocytes lose their ability to control glutamate level, a key chemical for the communication between brain cells. As a result, patients with Alexander's disease have an increased quantity of glutamate in the brain, which could explain the epilepsy observed in the patients. The high amounts of glutamate increase the development of protein tangles in the brain and brain inflammation. This protein, known as Tau, is involved in the communication between the cells. The inflammation also increases Tau impairment. This study will investigate the astrocytes role in the development of Tau protein tangles in Alexander's disease patients and its relation to the development of brain inflammation.