

How did we figure this out? Let's walk through the process!



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Three Genetic Mutations of the OXTR Gene Linked to Autism

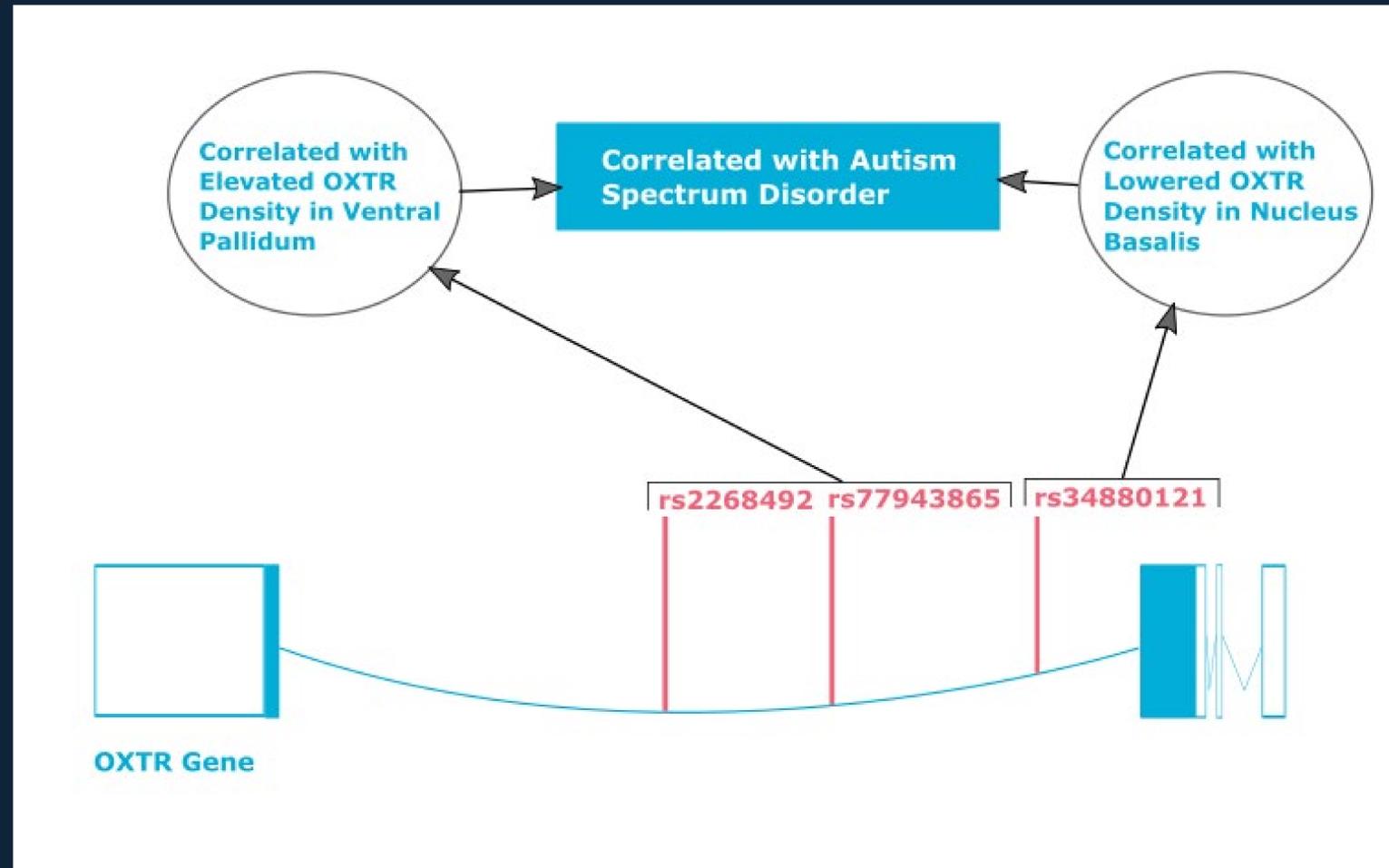
Introduction

In 2018, Dr. Sara Freeman published research showing a link between oxytocin receptor (OXTR) density in two brain regions: the Nucleus Basalis of Meynert (NBM) and Ventral Pallidum (VP) with autism.

This study takes the next step in examining the causes of autism by studying the relationship between genetic mutations (SNPs) in the OXTR gene and OXTR binding density in the VP and NBM.

Methods

An Illumina Infinium SNP assay was used to process DNA and identify SNPs. The raw data was clustered using GenomeStudio© software. Following this, the data was visualized using UMAP and a multivariable regression model was created to identify SNPs that have a significant relationship with OXTR binding density in the VP and NBM.



This image summarizes our research findings. OXTR stands for oxytocin receptor. The genetic mutations are marked in pink. Each of them are intron variants of the OXTR gene. My findings expand on an earlier study by Dr. Sara Freeman on OXTR density in the brains of autistic individuals.

Results

The UMAP visualization showed some clustering with the factors we had, but not all the variance was explained. This was also demonstrated in our regression results, which showed that the three SNPs in the center image (as well as age for the NBM) had significant relationships with OXTR binding in the NBM and VP. For both the NBM and the VP, the factors we had explained about 50% of the variation we're seeing, which is a good step forward, but more research is still needed.

What's Next?

A GWAS study could help us to account for the remaining variance by taking a broader view of the genome.

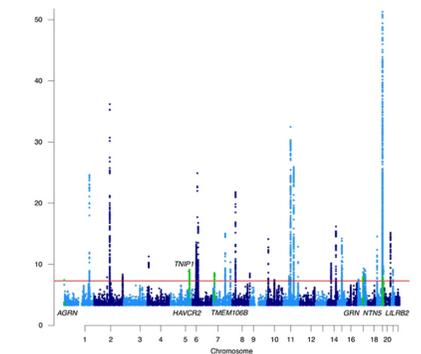


Image of an example GWAS study

Additionally, an RNA-Seq study using RNAScope would help us verify the relationship between OXTR gene expression and OXTR protein binding density.

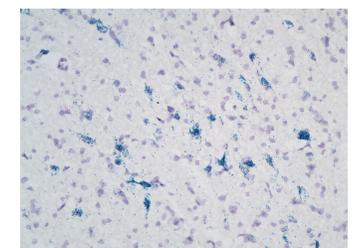


Image of an example RNAScope study

We plan to perform both of these studies in the coming months.

