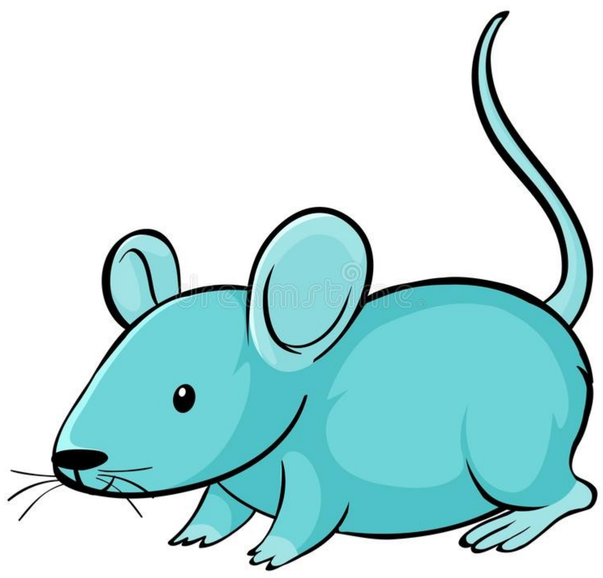
early brain development disrupted by 16p11.2 copy number variants in autism

Introduction

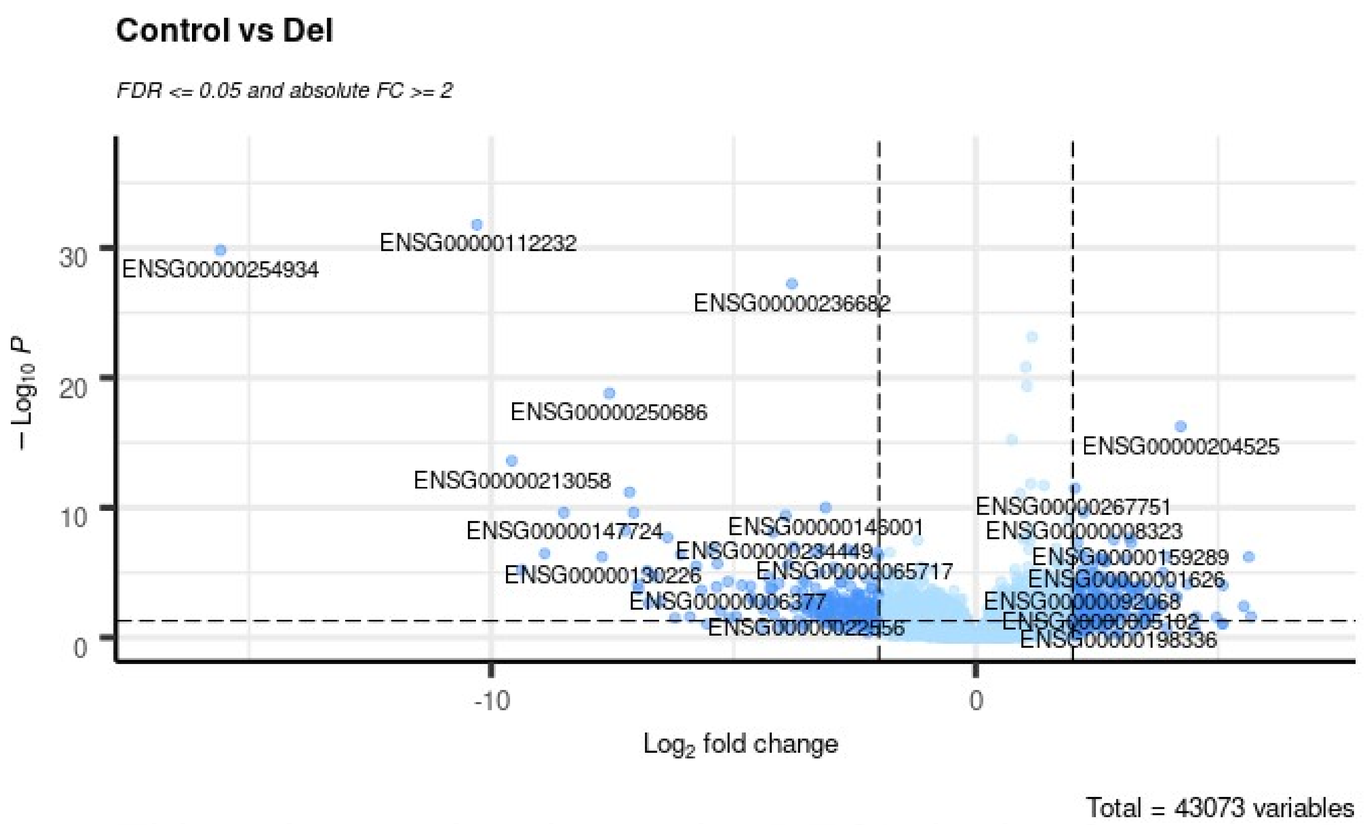
Insertions or deletions of the 16p11.2 region are often associated with neurodevelopmental disorders. Deletion of this region often leads to the autism spectrum disorder. A study [1] experimented with mouses that had extreme head sizes. This project uses the data from the study. The goal of this project is to see if the genes that are measured are different throughout each group (Control, Deletion and Duplication)

Conclusion

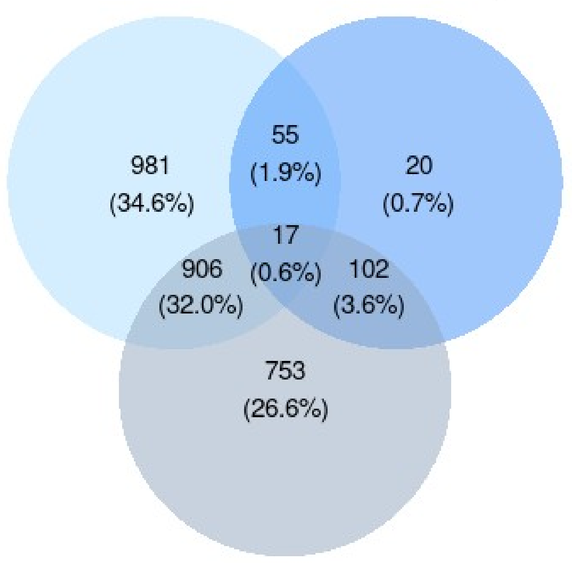
The article concluded that brain organoids faithfully recapitulate fetal development at the transcriptional level. There was no way i could conclude this too, the results simply do not give enough information to conclude that. What i could say is that since there's way more DEG's in the Deletion part, it is probably associated with ASD.



Results



This vulcano plot shows the DEG's in the Control vs Del. This is the comparison which has the most DEG's. Which makes sense, because it's the control group vs the group which is (probably) associated with ASD.



This venn plot compares the DEG'S from each group and vizualizes it nicely.

The plot shows that the control Del, and Del Dup easily have the most DEG'S (93.2%).

Materials and methods

Male mouses with the 16p11.2 region were tested to see if the region had effects on them. The study included six mouses with extreme head size (result of the region), and 3 healthy unrelated individuals as control. There were a total of 108 samples. 4 * 3 samples for each of the 3 groups, and 3 timestamps. IPSC, 1 month and 3 months.
The fold-change value was kept to 0.5.
All statistical tests were made with DESEq2.

References

[1] Cortical organoids model early brain development disrupted by 16p11.2 copy number variants in autism https://www.nature.com/articles/s41380-021-01243-6

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