Project proposal

The correlation between Schizophrenia and Epilepsy due to deletions on the *CYFIP1* gene

For this project the research "Reduced CYFIP1 in human neural progenitors as 15q11.2 deletion model: donor specific dysregulation of schizophrenia/epilepsy genes" (1) was chosen. It is stated in the publication that deletions occurring on a certain region on chromosome 15 (15q11.2, to be exact) increase schizophrenia and epilepsy risk, but that only some of the carriers of this mutation have either disorder. The purpose of this research was to investigate the role of a gene in the region mentioned above. The name of the gene is CYFIP1. The RNA-seq libraries were prepared using an Illumina TrueSeq RNA prep kit. The molecule that was extracted was polyA RNA and the total RNA was isolated from cells by using miRNAeasy Kit (Qiagen) which was done according to the manufacturer's protocol. Additionally, a DNaseI digestion step was also performed which was to ensure that samples were not contaminated with genomic DNA. To check the quality of the RNA it was assessed via an Agilent bioanalyzer.

To summarize the experiment, what was noticed is how the deletions in a certain region would not affect every individual, and in fact, this deletion is even common in the general population, occurring at a frequency of roughly 1/500. The difference that would result in whether a person would have either schizophrenia or epilepsy had to do with the actual expression of the gene compared to the locations of the deletions. What happens in a person when they get either of the disorders is that there is a reduced expression of the, already mentioned, 'CYFIP1' gene.

For this experiment RNA-seq was performed on NS (Neural Stem) and CYFIP1 NPCs (Neural progenitor cells) which were prepared from three different iPSC (Induced pluripotent stem cells) lines (these are in the data referred to as C2, C4 and C5).

What I wanted to do for this project is to try and remake several of he graphs found in the citation. There were many of those so I do not expect to be able to cover all of them. So as for now, I am still trying to understand everything better and then decide on which to recreate.

Neurodevelopmental disorders (NDDs)
Induced pluripotent stem cell (iPSC)
Neural stem/progenitor cells (**NS/NPCs)**

(1) https://www.ncbi.nlm.nih.gov/geo/query/acc.cgi?acc=GSE70935