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Project Title: Investigating the Role of the X Chromosome in Autoimmune Diseases Using Single-Cell Genomics

1. Introduction

During my internship at the Donnelly Centre for Cellular and Biomolecular Research, I focused on a project that investigates the role of the X chromosome in autoimmune diseases using single-cell genomics. The key objective of my research was to identify differentially expressed genes (DEGs) on the X chromosome between control and disease conditions in autoimmune diseases. This research aimed to enhance the understanding of the X chromosome's involvement in autoimmune conditions, potentially leading to the identification of novel therapeutic targets.

2. Materials and Methodology

To accomplish the research objectives, I employed various bioinformatics tools and techniques. The primary dataset I worked with was single-cell RNA sequencing (scRNA-seq) data, which allowed for the analysis of gene expression at the single-cell level. I used R programming for data processing and analysis, including the identification of DEGs using Seurat tool. The research was conducted in two main phases: data preprocessing and quality control, followed by differential gene expression analysis focused on the X chromosome.

3. Results and Discussion

The analysis revealed several X chromosome-related genes that were differentially expressed between the control and disease states in Crohn's disease. These findings suggest a potential role of the X chromosome in the pathogenesis of autoimmune diseases, aligning with previous research indicating a gender bias in autoimmune conditions. The implications of these results are significant, as they could pave the way for targeted therapies that specifically address the genetic components of autoimmune diseases linked to the X chromosome. However, further research is required to validate these findings and explore their clinical relevance.

4. Skills and Knowledge Gained

Throughout the internship, I acquired a range of technical skills, including proficiency in R programming and single-cell RNA sequencing data analysis. I learned how to effectively search for and interpret gene expression data, as well as how to critically read and comprehend complex scientific research. This experience also fostered my ability for self-directed learning, enabling me to independently acquire new knowledge and skills.

Additionally, I enhanced my understanding of autoimmune diseases and the role of the X chromosome in their development. Beyond these technical skills, I developed important soft skills such as communication, particularly in presenting complex research findings in a simplified and accessible manner. I also honed my teamwork abilities through close collaboration with other researchers and strengthened my problem-solving skills by successfully addressing the challenges encountered during data analysis.

5. Challenges and Solutions

One of the significant challenges I faced during the project was the complexity of single-cell RNA sequencing data. The inherent variability and high dimensionality of the data presented difficulties in analyzing and interpreting the results. To address this, I focused on leveraging advanced analytical techniques that could accommodate the complexities of the data, ensuring that my analysis was as robust and insightful as possible.

Another challenge was the limited time available to work on multiple datasets. Due to these time constraints, I had to prioritize my analysis by focusing on only two key datasets. This decision allowed me to concentrate on producing high-quality results rather than spreading my efforts too thin across multiple datasets.

Additionally, towards the last month of my internship, I encountered a major challenge when the data for my initial project became unavailable. This unexpected setback forced me to pivot to a new project with entirely new concepts. Despite the time pressure and the need to quickly adapt to new methodologies, I successfully overcame this challenge by rapidly acquiring the necessary

knowledge and skills to complete the new project. This experience taught me the importance of flexibility and resilience in research, as well as the value of being able to adapt to unforeseen circumstances.

6. Conclusion

My internship experience at the Donnelly Centre was highly enriching and significantly influenced my career goals. It provided me with a deeper understanding of the intersection between genetics and autoimmune diseases, reinforcing my interest in pursuing a career in bioinformatics and health informatics. The key takeaways from this experience include the importance of meticulous data analysis and the potential of genomics in uncovering new insights into complex diseases. Moving forward, I aspire to continue exploring the role of genetics in disease and contribute to the development of personalized medicine.

7. Acknowledgments

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