Delineating Expressional Difference in the Blood Mononuclear Cells between Healthy and Turner Syndrome Individuals

Romana Ishrat[1], Anam Farooqui[1], Naaila Tamkeen[2], Safia Tazyeen[1], and Sher Ali[3]

[1]Jamia Millia Islamia
[2]Jamia Millia Islamia Faculty of Natural Sciences
[3]Era University

March 22, 2023

Abstract

Turner Syndrome (TS) is a rare disorder associated either with complete or partial loss of one X chromosome in women. Comparing the healthy and individuals with Turner Syndrome may help elucidate the mechanisms involved in TS pathophysiology. Gene expression differences between healthy and individuals with Turner Syndrome were characterized using the systems-biology approach of weighted gene coexpression network analysis (WGCNA) on 182 microarray peripheral mononuclear blood samples (PBMC). The co-expression networks of healthy and TS had scale-free topology that ensures network robustness. In the process, five modules were preserved between healthy and TS, which carry several genes common in each module. Previously reported genes of TS, specifically, *PTPN22*, *RPS4X*, *CSF2RA*, and *TIMP1* were missing in their respective modules. Dysfunction, differential expression, or absence of these genes could lead to a progressive disruption of molecular pathways leading to the pathophysiology of TS. Indeed, we observed a significant difference in the functions of these modules when compared within and across the healthy and TS samples. We identified 4 clusters in the PPI network constructed from the top 15 K $_{\text{ME}}$ enriched in significant functions. Overall, our work highlights the potential molecular functions, pathways, and molecular targets of TS that can be exploited therapeutically in the human health care system.

Hosted file

`Manuscript.docx` available at https://authorea.com/users/598468/articles/631027-delineating-expressional-difference-in-the-blood-mononuclear-cells-between-healthy-and-turner-syndrome-individuals

Hosted file

`Figure 1.eps` available at https://authorea.com/users/598468/articles/631027-delineating-expressional-difference-in-the-blood-mononuclear-cells-between-healthy-and-turner-syndrome-individuals