Studying the Effects of the Missing X Chromosome on the Liver

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The purpose of this retrospective and observational study was to see the effect of the missing X chromosome in women (a condition known as Turner Syndrome) on the liver. It was expected that the missing X chromosome would deteriorate liver function and therefore increase liver enzymes. The method followed was the obtaining and analysis of patient's liver enzymes, karyotype, and other relevant information. Six out of twenty-three females with Turner Syndrome had high liver enzymes (> 50 U/L) (Alanine Aminotransferase (ALT), Asparate Aminotransferase (AST), or both). This is about three times as high as the percentage of females with high liver enzymes in the control group. Additionally, the degree of elevation of the liver enzymes in patients with Turner Syndrome (over all BMI ranges) tended to be higher than those without the missing X chromosome. One third of the Turner Syndrome patients who showed high liver enzymes were mosaic (not all of their cells were missing the X chromosome), implying that there is not a correlation between karyotype and liver involvement. Older patients tended to have higher liver enzymes, as expected. The next step to expand upon this research would be to study whether increased liver enzymes in patients with Turner Syndrome progresses to future complications such as cirrhosis of the liver. Additionally, one can study ways to improve the livers of Turner Syndrome patients; for example, ursodeoxycholic acid may be a way to prevent liver deterioration over time.

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