Nicholas Fehrer
01195688
Genetics Writing Assignment 3
Professor Rinehart
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The article I chose for writing assignment 3, *The Genetic and Clinical Significance of Fetal Hemoglobin Expression in Sickle Cell Disease*, is a genetics related article which can be found in the peer reviewed journal, Krager. The article mostly focuses on the sickle cell disease and fetal hemoglobin expression. Since sickle cell disease is heterogeneous and is a genetic disease where the red blood cells form to the shape of a sickle and die earlier than intended leaving the body with a deficiency. The hemoglobin is a tetrameric protein within the red blood cells that carries oxygen through the body to the various organs and tissues and brings carbon dioxide into the lungs of an individual. The globin contains elements that are crucial to transcription and the regulatory elements within the body making them a large part of not only oxygen transportation but also gene regulation. The main interest the researchers had with fetal hemoglobin though, is how they switch from embryonic to adult life, for instance the HbF is predominant at birth but declines to almost nonexistent six months after the child is born. With all this background information stated, and with the way fetal hemoglobin work, we can finally dive into the study itself. Infants are asymptomatic for the sickle cell disease until they are around 6 months old, and studies have shown that HbF enhances the process. Analyzing these studies show that patients with sickle cell disease who had higher HbF had fewer problems and that any percentage less than or equal to twenty continually showed a lesser risk of a 'major clinical event,' given to us from data recorded mainly from the Middle East. Kuwait was one of the most researched countries where the frequency of HbF is higher at almost twenty-two percent, a perfect place to study the phenotype. Within the ten-year study the most reported symptom of hospitalized patients was pain, gallstones, specifically located in the head, and silent brain infracts. With some further research they found that neurocognitive function in the children affected with the silent brain infracts was comparable to their siblings which was quite surprising. Many residents in the study had a HbSS phenotype which leaves the door open to mutation to the Sb0, leading to the rise in pain over time. With higher levels of HbF now than before the mild phenotype has been able to show no direct connection between the HbF and the other clinical phenotypes. With higher HbF levels consisting of much of the populations, the total number of F-cells is the most important part in determining the role of the HbF. Therefore, the proportion of these cells that have enough HbF to stop polymerization, being the most effect way to understand and find the severity of the sickle cell disease. The author concluded that there is much evidence leading toward HbF improving the consequences of the sickle cell disease, being shown within the Kuwaiti population throughout the research. Although the report and research are quite extensive all the facts are yet to be truly understood including haplotype-specific genetic factors and ethnocentrisms that have yet to be even found.

Adekile Adekunle. The Genetic and Clinical Significance of Fetal Hemoglobin Expression in Sickle Cell Disease. *Medical principles and practice: international journal of the Kuwait University, Health Science Centre; https://doi.org/10.1159/000511342 (2021).*