Analysis of the HBG2-4kb DNase I Hypersensitive Site Before and After the Knockout of the HBD-1kb Region in the Beta-globin Locus

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There are a wide variety of Beta-globin related disorders including sickle cell disease and Beta-thalassemia. There are five main genes in the Beta-globin locus: epsilon, gamma two, gamma one, delta, and beta. Each corresponds to expression at a certain developmental stage in the life of a human. It is known that the knockout (KO) of the HBD-1kb (Hemoglobin Delta) region leads to the decrease in accessibility of the HBG (Hemoglobin Gamma) and HBD promoters, as well as, most of the Locus Control Region's (LCR) hypersensitive sites (HS) in the Beta-globin locus. However, an HS site not accounted for is the HBG2-4kb enhancer region. Therefore, the purpose of this study is to use bioinformatics to analyze the effects HBD-1kb KO has on the activity of HBG2-4kb. In this study, the datasets were gathered from the In Situ Capture of Chromatin Interactions by Biotinylated dCas9 study and converted into bigWig files so they could be visualized. After the study was completed, the data showed that the HBG2-4kb enhancer region decreased by approximately a factor of 10.855, which makes the KO to control ratio approximately 0.092. From this information, one can conclude that these two regions interact with one another, and the HBD-1kb region in important for the proper functioning of the HBG2-4kb enhancer site. This information, along with future research in the Beta-globin locus, will allow researchers to better understand at a molecular level, the Beta-globin locus, the Beta-globin related disorders, and the possible treatments and cures for these disorders.