

DESCRIPTION FOR THE GENERAL PUBLIC (IN ENGLISH)

Substantial progress has been made in the research into the causes of inherited human diseases, largely thanks to the expansion of knowledge regarding the human genome, the discovery of new markers of genetic variability, and advancements in genomic analysis technology. Additionally, the development of novel bioinformatics tools has aided in the analysis of the vast amount of data generated. Currently, the focus of genomic research is shifting towards establishing the molecular relationship between mutations and diseases. Changes in genomic sequence can impact various interacting networks that collectively influence susceptibility to complex traits, affecting chromatin structure, gene expression, and localization. This proposal concentrates on studying the molecular basis of a hereditary limb disorder known as Clubfoot. Since this disease is multifactorial in nature, the hypothesis is that different gene defects or regions could lead to the same patient phenotype. The main objective of the study is to establish the genotype-phenotype correlation in Clubfoot disease. The research will focus on functional analysis of the identified mutation in a family with Clubfoot and determining the significance of these changes in the etiology of Clubfoot.