Using Comparative Genomic Hybridization to Identify Unique Mutation in Duchenne Muscular Dystrophy Patients in Saudi Arabia

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Duchenne Muscular Dystrophy (DMD) is an inherited disorder that involves muscle weakness and occurs in approximately 1 out of every 3,600 male infants. The average life expectancy for individuals afflicted with DMD is 25 years old. It is caused by a defective gene in the dystrophin protein. The aim of this novel study is to identify unique mutations in the dystrophin gene causing DMD in Saudi patients and carriers, using comparative genomic hybridization (CGH). CGH can be used to overcome multiplex PCR limitations to identify unique mutations in the dystrophin gene in Saudi Arabian DMD population. In this experiment, DNA from clinically diagnosed DMD anonymous patients was extracted from blood samples. Following, the DNA was fragmented using restriction endonucleases, amplified and labeled with phlorophores, and then purified using affinity columns. The next step was to hybridize the samples to labeled reference DNA, washed them and scan them to identify applicable mutations. As a result, this experiment identified deletion mutations within the dystrophin gene in Saudi DMD patients. In one case specifically, the protocol successfully identified a deletion mutilation between exons 45 and 52 confirming the disease, while it questioned the diagnosis of another by not identifying any mutations at all in the dystrophin gene. Thus, CGH shows more accurate results than other techniques and overcome the gold standard multiplex PCR limitations. Potentially this technique can allow for the identification of mutations in the dystrophin gene and the design of its specific gene therapy.

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