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Thesis Title	***Molecular Characterization Of Dystrophin Gene In A Sample Of Iraqi Patients With Muscular Dystrophy***			
Year	**2013**			
Abstract	**Background: Dystrophinopathies are the commonest forms of muscular dystrophy and comprise clinically recognized forms, Duchenne Muscular Dystrophy (DMD), and Becker Muscular Dystrophy (BMD). Mutations in the dystrophin gene which consist of large gene deletions (65%), duplications (5%) and point mutations (30%) are responsible for reducing the amount of functional dystrophin protein in skeletal muscle fibers leading to fiber destruction and disease.** **Aim: The aim of this study was to investigate the rate, and distribution of deletions in 10 exons of Dystrophin gene in a group of Iraqi dystrophinopathy patients using the multiplex polymerase chain reaction (MPCR). In Iraq, no previous work has been reported on the mutation patterns of Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD). This study concentrate mainly at the spectrum of deletions in the 'distal hot spot' region of the DMD/BMD gene in Iraqi DMD/BMD patients .** **Patients materials and methods: It is a case control prospective study which include 27 clinically diagnosed DMD/BMD patients and six suspected carriers. A written consent was obtained from each family for going the research as well as ethical committee approval. Forty six apparently healthy individual were included as a control group. Clinical diagnosis was based on physical examination, progressive muscle weakness, and muscle strength, high level of serum creatine phosphokinase (CPK), myopathic changes on electromyography (EMG) and family history of other affected individuals in the family. Blood samples were collected in 5-6 ml EDTA tubes by venepuncture. The DNA was extracted by using the Wizard Genomic purification kit (Promega/USA) and the quantity was estimated by UV-absorbance.**			

	Ten exons of the dystrophin gene were examined (19 ,45 ,46 ,47 ,48 ,49 ,50 ,51 ,52,53) using synthesized primers with complementary sequences and set in five different multiplex PCR groups. Each multiplex PCR group amplified a total of two exons. These primers detect the deletions in the given exons in patients and suspected carriers. Normal controls were included for validation. Then the products of PCR amplifications were subjected to electrophoresis and visualized by UV- light system. **Results:The rate (relative frequency) of subjects with any positive exonal deletion { among the10 selected and tested exons was significantly higher (85.2%) among patients compared to that among the suspected carriers (33.3%)} .The distribution of exonal deletions among patients compared to suspected carriers were statistically significant. The frequency of deletions detected in male patients (~82%) was higher than frequencies mentioned in the other studies of comparison. The control group show no deletion in all tested exons.** **Conclusions: Multiplex PCR technology was utilized to demonstrate the frequency of 10 exons deletions in a limited group of Iraqi DMD/BMD patients. The overall distribution of deletion mutations in the distal 'hot spot' region was higher than that of DMD/BMD cases investigated elsewhere. The study also serves as a good starting point for further investigations into the genetic aspects of the Iraqi DMD/BMD population.**