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Research Details :

Research Title : *Molecular Diagnosis of Duchenne Muscular Dystrophy*
التشخيص الجزيئي لمرض دوشان لأضمحلال العضلات

Descriptipn : Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder that affects muscle and causes progressive weakness as a result of the production of non-functioning dystrophin protein or no dystrophin produced at all. Dystrophin, the product of the dystrophin gene, is one of several membrane proteins that form the dystrophin-glycoprotein complex, which helps to maintain the integrity of muscle cells; loss of these proteins leads to the wasting of muscle cells and to the pathology of DMD. This study is conducted to look into the spectrum of DMD gene in Saudi patients with Duchenne. An accurate molecular diagnosis achieved in DMD/BMD patients has been helpful in advising the patients about the pattern of inheritance and determination of carrier status is crucial to a complete and comprehensive family profiling In this study, the dystrophin gene were analyzed in 8 DMD patients with elevated level of creatine kinase (CK) and their family members (25 subjects) for the presence of deletions by using four different multiplex PCR sets each amplifying a total of 6, 7, 7 and 6 different exons. Deletions were found in 0, 62.5, 62.5 and 37.5 % of DMD patients with each of the four sets, respectively. With all the four sets, the deletion rate was 62.5 % (5 of 8 patients). Sixty two percent of the deleted exons were located in the distal hot spot, none (0 %) to the proximal hot spot while 37 % were scattered over both. No deletion in DMD gene was detected in the control samples. This is the first study on DMD which has revealed that there was no deletion in the distal hot spot region.

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Researchers :

Researcher Name (Arabic)	Researcher Name (English)	Researcher Type	Degree	Email
عائشة بنت حسن عبدالله عليمي		Researcher	.	