

Summary

Duchenne Muscular Dystrophy (DMD) is a common genetic disease that affects approximately one in 3000 males. Becker Muscular Dystrophy (BMD) is less common, affecting approximately one in 30,000 males. Both diseases result from a mutation in the gene located at Xp₂₁ which encodes dystrophin, a sarcolemmal protein abundant in skeletal and cardiac muscle cells. Dystrophin is typically absent in DMD and reduced or abnormal in size in BMD. DMD typically is diagnosed between the ages of 3 and 7 years and is characterized by progressive skeletal muscle weakness with loss of ambulation between the ages of 7 and 13 years. The BMD phenotype is clinically more heterogeneous with initial presentation in the teenage years.

Approximately 70% of boys with Duchenne/Becker Muscular Dystrophy (DBMD) have mothers who carry an Xp₂₁ mutation; the remaining boys have a new occurrence of the disorder attributable to a spontaneous mutation. Most female carriers do not manifest neuromuscular problems, but it is well established that some are more likely to develop Dilated Cardiomyopathy (DCM), even at a young age.

We performed this study to evaluate the usefulness of cardiac troponin I, in the assessment of cardiac affection in patients with dystrophies.

This study was conducted on 30 Egyptian male patients with muscular dystrophies, of age range of 4-18 years. The participants were divided into two groups:

- Group (I) of 30 male patients.
- Group (II) of 20 normal control male participants.

After review of medical records, these patients will be subjected for:

- Thorough history and physical examination.
- Radiological and laboratory investigations:
 - **Routine:** Complete blood count, biochemistry, chest X-ray, and 12 leads electrocardiogram.
 - **Specific:** Echocardiography to assess cardiac function, Creatin Kinase MB (CK-MB) assays.
 - **Cardiac troponin I measurement** by monoclonal anti body-based diagnostic immunoassay.

Our study showed no significant difference between the patients and control groups regarding age. It showed highly significant rise in both serum cTnI and CK-MB.

We showed highly significant positive correlation cTnI and CK-MB while a negative correlation between it and EF. Our study revealed that the patients on wheel chair presented higher levels of cTnI.

Also, there was a highly significant negative correlation between cTnI and EF while a positive correlation between it and LVEDD and LVESD, AO, LA, RV, IVS and LVPW.

Our thesis study showed highly significant decline in EF in patients group in comparison to control group yet still the cardiac function is within the normal range. Our study showed significant decline in the motor function of the patients with 40% were on wheel chair.

Also, there was a highly significant negative correlation between CK-MB and EF while a positive correlation between it and LVEDD and LVESD.
