Dystrophin protein

- Maintains muscle membranes
- Connects intracellular actin to transmembrane proteins
- Binds α- and β-dystroglycan in membrane
- found in cardiac and smooth muscle and in some brain neurons
- Its large size may make it more prone to sporadic mutations
- Its gene on X chromosome (Xp21)
- largest human genes

Loss of dystrophin protein cause

- myonecrosis
- Creatine kinase elevation
- Muscle replaced with fat/connective tissue
- Muscle Weakness , difficult to run or climb ,Need Wheelchair
- Heart will be affected
- Mental retardation may occur

Duchenne Muscular Dystrophy:

- More common More severe
- X-linked disease
- show complete absence of dystrophin gene (deletion)
- Affected boys normal first few years (1 per 3500)
- Weakness develops age 3-5
- Wheelchair usually by age 12
- Death usually by age 20
- Diagnosis: Elevated Creatinin Kinase and genetic Study

Sign and symptoms:

- Clumsiness and an inability to keep up with peers
- Difficulty running, jumping, climbing stairs
- Use hands to push themselves up from chair (Gower's sign)
- Muscle replaced with fat/connective tissue Calf enlargement "Pseudohypertrophy"
- Heart failure and arrhythmias
- mental retardation
- death due to Respiratory insufficiency / pneumonia / cardiac decompensation

Becker Muscular Dystrophy:

- X-linked disease
- have point mutations and make residual of dystrophin gene
- Milder than Duchenne

- Late age of onset and Becomes symptomatic later
- Many patients live well into adulthood and have a nearly normal life span.
- Cardiac involvement may be the dominant clinical feature
- Diagnosis: Elevated Creatinin Kinase and genetic Study

Emery-Dreifuss muscular dystrophy (EMD):

- caused by mutations affecting structural proteins found in the nucleus.
- heterogeneous: disorder X-linked and autosomal dominant form
- X-linked form results pf mutations in the gene encoding the protein emerin
- autosomal dominant form is caused by mutations in the gene encoding lamin
- defects in these proteins compromise the structural integrity of the nucleus in cells
- These proteins may also regulate chromatin structure and there by affect gene expression patterns
- progressive muscle weakness and wasting, contractures of the elbows and ankles, and cardiac disease
- cardiac involvement is severe, lead to sudden death in up to 40% of patients