- A 62-year-old lady has attended the emergency department with several days of shoulder weakness and pain. There are no abnormalities on plain X-rays but the emergency department doctor was concerned about a purplish discolouration around the eyes and prominent nail fold telangiectasia which the patient has not really noticed before.
- **Which of the following would you do next in this patient?**
- Serum creatine kinase
- Serum erythrocyte sedimentation rate
- Analgesia and physiotherapy
- Prednisolone 1 mg/kg/day
- Thorough clinical examination

Myopathies

Muhammad Ishfaq

Diseases of Muscle

- Hereditary or Acquired.
- Proximal muscle weakness.
- Cardiorespiratory involvement, family history, drugs exposure.
- EMG and muscle biopsy.

Hereditary

1. Muscular dystrophies,
2. Muscle channelopathies,
3. Metabolic myopathies (including mitochondrial diseases)
4. And congenital myopathies.

Acquired

- These include the
- inflammatory myopathies, or
- myopathy associated with a range of metabolic and endocrine disorders or drug and toxin exposure

Muscular Dystrophies

- Myotonic is most common. Duchene and Becker are other
- progressive muscle destruction and may be associated with cardiac and/or respiratory involvement and sometimes non-myopathic features.
- Onset is often in childhood, although some patients, especially those with myotonic dystrophy, may present as adults.
- Wasting and weakness are usually symmetrical, without fasciculation or sensory loss, and tendon reflexes are usually preserved until a late stage. Weakness is usually proximal, except in myotonic dystrophy type 1, when it is distal

Inv

- Genetic testing
- EMG
- Muscle biopsy
- CPK elevation in Duchene and becker,
- Echo and ECG for conduction defects.

- No specific treatment.
- Steroids in Duchene.

Inherited Metabolic Myopathies

- There are a large number of rare inherited disorders that interfere with the biochemical pathways that maintain them energy supply (adenosine triphosphate, ATP) to muscles. These are mostly recessively inherited deficiencies in the enzymes necessary for glycogen or fatty acid (β-oxidation) metabolism.
- McArdle Disease,
- Acid Maltase deficiency, pompe's disease

Mitochondrial disorders

- vision (optic atrophy, retinitis pigmentosa, cataracts),
- hearing (sensorineural deafness) and
- the endocrine,
- cardiovascular,
- gastrointestinal and
- renal systems

- especially if there is evidence of maternal transmission.

- Mitochondrial dysfunction can be caused by alterations in either mitochondrial DNA or genes encoding for oxidative processes.

- Thus, patients with exercise intolerance, myalgia and sometimes recurrent myoglobinuria may have isolated pathogenic mutations in genes encoding for oxidation pathways

- Many of these mitochondrial disorders are inherited via the mitochondrial genome, down the maternal line

- Diagnosis is based on clinical appearances, supported by muscle biopsy appearance (usually with ‘ragged red’ and/or cytochrome oxidase-negative fibres), and specific mutations either on blood or, more reliably, muscle testing. Mutations may be due either to point mutations or to deletions of mitochondrial DNA

- MERRF, MELAS, leber, kearn sayer,

Channelopathies

- Inherited abnormalities of the sodium, calcium and chloride ion channels in striated muscle produce various syndromes of
- familial periodic paralysis,
- myotonia and
- malignant hyperthermia, which may be recognised by their clinical characteristics and potassium abnormalities. Genetic testing is available.

Inflammatory

- Polymyositis
- Dermatomyositis (muscle disease plus skin involvement)
- Inclusion body myositis

- Elevation of CK
- Muscle biopsy

- Steroid and immunosuppressive therapy

Endocrine

- Hypothyroidism
- Hyperthyroidism
- Acromegaly
- Cushing’s syndrome (including iatrogenic)
- Addison’s disease
- Conn’s syndrome
- Osteomalacia

Drugs

- Glucocorticoids
- Statins
- Amiodarone
- β-blockers
- Opiates
- Chloroquine
- Ciclosporin
- Vincristine
- Clofibrate
- Zidovudine

Toxic

- Alcohol (chronic and acute syndromes)
- Amphetamines/cocaine/heroin
- Vitamin E
- Organophosphates
- Snake venoms