CHILD WITH ABNORMAL GAIT
PATIENT’S PROFILE

- NAME: Mohammad Ramzan
- AGE: 8 years
- SEX: Male
- RESIDENCE: Chakwal
- DATE OF ADMISSION: 23rd July 08
PRESENTING COMPLAINTS

- Frequent falls during walking 1-2 years
- Progressive difficulty in walking 3 months
PAST HISTORY
not significant

DRUG HISTORY
not significant

BIRTH HISTORY
SVD at hospital
no h/o birth anoxia
no h/o neonatal jaundice
FEEDING HISTORY
Current caloric intake is about 1500 cals/day

VACCINATION HISTORY
Completely immunized

DEVELOPMENTAL HISTORY
Milestones achieved at appropriate ages.
FAMILY HISTORY

3rd among the sibship of 5. no family history of any illness.

SOCIOECONOMIC HISTORY

Lower middle socioeconomic class.
EXAMINATION

GPE:
  Pulse: 88 beats/min
  RR: 24 breaths/min
  Temp: 98 F
  B.P: 90/60

ANTHROPOMETRY:
  Weight: 16 kg (below 5th centile)
  Height: 127 cm (on 50th centile)
  OFC: 52 cm (mean)

Rest of the general physical examination was unremarkable.
EXAMINATION

GAIT:

Toe walking, plantar flexion of both feet, hips extended and abducted, lateral twisting of the trunk & outstretched arms to maintain balance.
EXAMINATION

CNS:

HMF & Speech: Normal
Cranial nerves: intact
Sensory system: intact
Motor system:
  Tone increased.
  Power 5/5 in upper limbs & 4/5 in lower limbs.
  Reflexes exaggerated.
  Clonus negative.
  Plantars downgoing.
CEREBELLUM:  
Intact

SPINE:  
Normal

Rest of the systemic examination was unremarkable.
DIFFERENTIAL DIAGNOSIS

- DYSTONIA MUSCULORAM DEFORMANS
- WILSON’S DISEASE
- RAPID ONSET DYSTONIA- PARKINSONISM
INVESTIGATIONS

- **CBC**
  - Hb: 9.9 gm/dl
  - TLC: 9.46x10³ /mm³
  - PLTS: 321x10³ /mm³

- **RFT’s**
  - UREA: 4.1 mmol/l
  - CREATININE: 81 umol/l

- **SERUM ELECTROLYTES**
  - Na: 139 mmol/l
  - K: 3.68 mmol/l

- **LFT’s**
  - ALT: 29 U/l
INVESTIGATIONS

■ MUSCLE ENZYMES

- CK: 250 U/l
- LDH: 200 U/l

■ Serum ceruloplasmin: 300 mg/dl
WORK UP DONE

OPTHALMOLOGICAL EXAMINATION:
no KF ring

EMG / NCV:
Normal

CT SCAN BRAIN:
Normal
FINAL DIAGNOSIS

- DYSTONIA MUSCULORAM DEFORMANS
MANAGEMENT DONE

- Tab L-DOPA
- Physiotherapy
- Counselling
DEFINITION

Dystonia is a neurologic movement disorder characterized by sustained muscle contractions, usually producing twisting and repetitive movements or abnormal postures or positions.
CLASSIFICATION OF DYSTONIA

- Anatomical distribution
- Age at symptom onset
- Etiology
ACCORDING TO ANATOMICAL DISTRIBUTION

- Focal
- Segmental
- Multifocal
- Hemidystonia
- Generalized
ACCORDING TO AGE OF ONSET

Childhood onset---- 0 to 12 years.

Adolescent onset---- 13 to 20 years.

Adult onset---- older than 20 years.

In broader terms:
Early onset---- before 21 years.

Late onset---- later than 20 years or in late twenties.
ACCORDING TO ETIOLOGY

- Primary (idiopathic) dystonia.
- Secondary dystonia.
- Dystonia-plus syndromes.
- Heredodegenerative dystonia.
Signs Potentially Suggestive of Secondary Dystonia

- Patient history of a possible causative factor.
- Associated neurologic signs.
- Dystonia that occurs during periods of rest.
- Initial leg involvement during adulthood.
- Hemidystonia.
- Early onset of speech disturbance.
- Abnormal laboratory test results.
- Abnormal neuroimaging results.
- Signs suggestive of psychogenic causes.
Primary (Idiopathic) Dystonia

- Often referred to as "idiopathic torsion dystonia" or ITD

- Three main types:
  - DYT1 dystonia.
  - DYT6 dystonia.
  - DYT7 dystonia.
Dystonias Caused by *DYT1* Gene Mutation

- Also known as Dystonia musculorum deformans (DMD) or Oppenheim dystonia
- Mean age of onset of *DYT1* dystonia is 8-9 years.
- In 90 to 95 % of cases, symptoms begin in a leg or an arm with secondary generalization.
- Rate of progression is extremely variable.
For patients with initial arm involvement

- Dystonic spasms first appear when performing specific activities.
- With progression spasms occur with other unrelated activities of the arm-overflow phenomenon.
- Eventually, dystonia may be present at rest.
- 50% chance of progression to generalized dystonia.
Patients with initial leg involvement

- Distinctive gait abnormalities.
- The hip may be extended or abducted with plantar flexion of feet, inward turning of heels & extension of knee.
- In addition, severe flexion of the trunk may result in a "bent over" posture with extension of the neck.
- Eventually, symptoms are present even at rest.
- 90% chance of progression to generalized dystonia.
PATHOPHYSIOLOGY:

- Autosomal dominant disorder.
- Mutations in DYT1 gene which is mapped on chromosome 9.
- 30 to 40 % develop symptoms due to variable expressivity & low penetrance.
- Regulates production of a protein known as TORSIN-A
DIAGNOSIS

- A thorough patient history
- A careful family history
- Examination
  - Ophtamology
  - Neurological
DIAGNOSIS

- Laboratory studies,
  - Blood and urine tests
  - Analysis of cerebrospinal fluid (CSF)
  - Erythrocyte sedimentation rate (ESR)
  - Antinuclear antibodies (ANA) studies
  - Tests to detect the presence of acanthocytes
  - Serum ceruloplasmin
- EMG/NCV
- Neuroimaging studies
There are three main approaches to the treatment of dystonia:

- Oral medications
- Injections of therapeutic agents directly into dystonic muscle
- Surgery
MEDICATIONS USED

- **Benzodiazepines**
  Diazepam and clonazepam are commonly used

- **Baclofen**
  Administered orally or directly to the spinal cord

- **Anticholinergics**
  Trihexyphenidyl & diphenhydramin are commonly used

- **Dopamine-blocking or dopamine-depleting agents**
  Tetrabenazine is the most widely

- **Neuroleptic drugs**
  Clozapine and olanzapine
DRUGS USED FOR DIRECT INJECTIONS

- BOTULISM TOXIN A
- BOTULISM TOXIN B

...are commonly used for direct injection into the affected muscles...

- MODE OF ACTION

Blocks the release of acetylcholine, thus relaxes the muscle
Surgical Treatment of Dystonia

- Thalamotomy and Pallidotomy
- Deep Brain Stimulation
- Myectomy (Myotomy)
- Ramisectomy and Rhizotomaty
- Peripheral Denervation
THANK YOU