Case presentation

Dr Rupa

1st year pg

In paediatrics department

- ***XYZ,*** 6yr boy
- From Nalgonda

Chief complaints

- Weakness of both lower limbs- 1yr
- Difficulty in standing from sitting position - 6months.

History of presenting illness

- The child was able to walk and run till 5 years of age. Later, he developed insidious onset of difficulty in standing from sitting position, which was gradually progressive. Then, he had frequent falls and difficulty in walking in upstairs.

- No h/o myalgia, rash, contact tuberculosis, recurrent RTI or choking episodes.
- Antenatal: non consanguineous marriage, Regular ANCs done, iron and folic acid taken.
- Birth: Delivered by LSCS, cried immediately after birth, birth weight 2.5kgs, no h/o NICU admission.

- Developmental history:

Neck holding -6mon

Sitting with support-1yrs

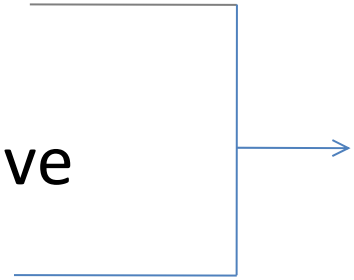
Standing with support-2yrs

Walking without support-3yrs

Fine motor

Social &adaptive

Language



attained as per the age

- Immunized as per the schedule.
- Family history: no similar complaints in the family

- General physical examination:
  - No pallor
  - No icterus
  - No clubbing
  - No lymphadenopathy
  - No oedema
- Head to toe examination: Normal
  - No dysmorphic features
  - No neurocutaneous markers.
  - Hypertrophy of calf muscles

- CENTRAL NERVOUS SYSTEM EXAMINATION:

  Conscious, oriented to time, place, person.

  Speech normal.

  Cranial nerves: I to XII intact

- Sensory system :intact
- Motor system:

Bulk of muscle: Rt upper arm: 14cms

Rt forearm :13 cms

Rt mid thigh : 24cms

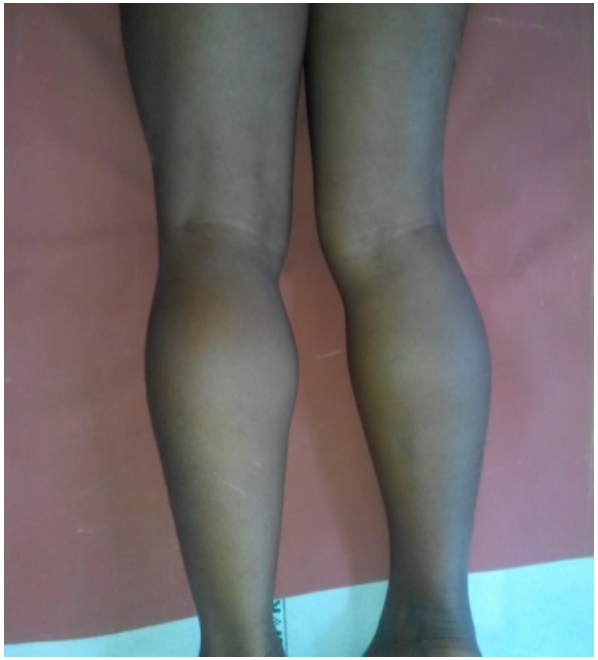
Rt calf muscle: 22cms

Lt upper arm:14 cms

Lt forearm :13cms

Lt mid thigh :24cms

Lt calf muscles :22.5cms



- Tone :normal in all four limbs
- Deep tendon reflexes : normal
- Plantar reflex: flexion response seen

Musculoskeletal examination:

Power :

	Rt	Lt
Deltoid	4/5	4/5
Biceps	4/5	4/5
Triceps	4/5	4/5
Brachioradialis	4/5	4/5
Flexors of forearm	4/5	4/5
Extensors	4/5	4/5
Hand muscles	5/5	5/5
Gastrocnemius	4/5	4/5
Gluteus maximus	4/5	4/5
Gluteus minimus	4/5	4/5
Calf muscles	3/5	3/5

- GAIT: waddling gait , with toe walking
- Gower sign: positive.
- All Joints, range of movements :Normal.
- Spine examination : Normal.

- Respiratory system:

  Bilateral air entry present.

  Normal vesicular breath sounds heard.
- Per abdomen examination:

  soft, no organomegaly.
- Cardiovascular system:

  S1,S2 Normal.

  no murmur.

Investigations

- Complete blood picture:

  Hb: 13.5 gm%

  Total count:11000

  N :40% L:55% E:3%
  M:2% B:0%

  Platelet count:2.8 lakh

  ESR:20

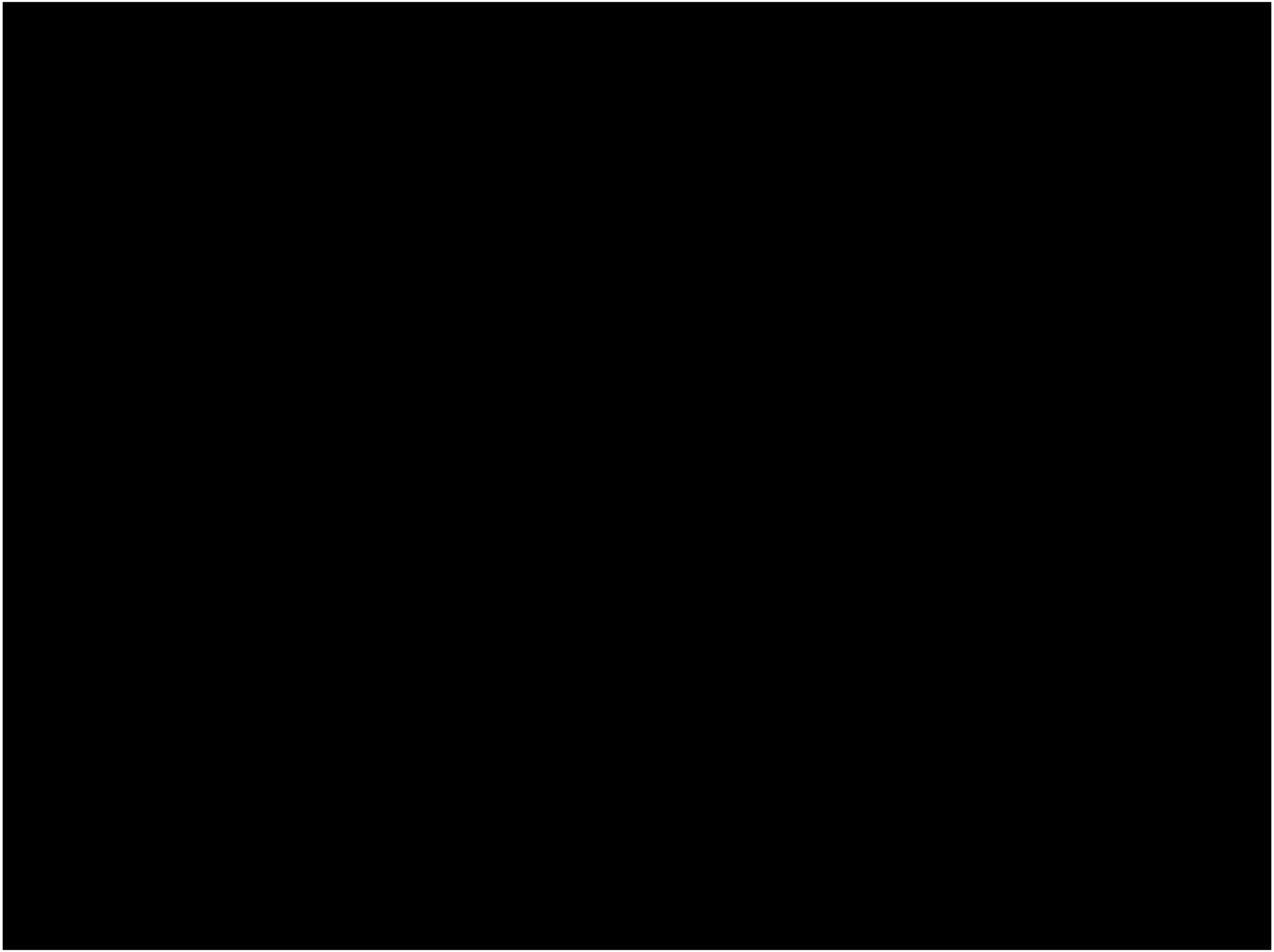
- Creatinine kinase level: 1870IU/L
- LFT, RFT, Serum electrolytes: with in normal limits
- Nerve conduction velocity: normal
- Molecular study for dystrophin gene: report awaiting
- Muscle biopsy: Not done.

SUMMARY

- 6 yr boy with progressive difficulty in walking, climbing upstairs, and frequent fall for 1yr
- O/E proximal muscle weakness, calf muscle hypertrophy, positive gower sign

Provisional diagnosis

- Progressive proximal muscular weakness
- ? Duchenne muscular dystrophy
- without cardiac or respiratory complications.



Case 2

- Xx 8yrs boy
- From miryalguda

Chief Complaints

- Difficulty to get up from sitting position- 2 yrs
- Difficulty in climbing upstairs – 1yr.
- Frequent falls – 6months

History of present illness:

The child was able to walk and run till 6 years of age. Later, he developed insidious onset of difficulty in standing from sitting position, which was gradually progressive. Then, he had frequent falls and difficulty in walking upstairs.

- No h/o myalgia, rash, contact TB, recurrent RTI, or chocking episodes.

- Antenatal : non consanguineous marriage, Regular antenatal checkups done. iron and folic acid taken.

- Birth :Delivered by LSCS, cried immediately after birth, birth weight 2.5kgs.
- No h/o NICU admission.

- Developmental history:

Neck holding -6mon

Sitting with support-1yrs

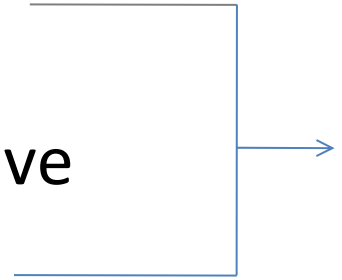
Standing with support-2yrs

Walking with support-$2^{1/2}$yrs

Fine motor

Social &adaptive

Language



attained as per the age

- Immunization done as per the schedule.

- Family history : h/o similar complaints to other sibling in the family

- General physical examination:
  - pallor present
  - No icterus
  - No clubbing
  - No lymphadenopathy
  - No oedema
- Head to toe examination: Normal
  - No dysmorphic features
  - No neurocutaneous markers

- CENTRAL NERVOUS SYSTEM EXAMINATION:

  conscious, oriented time, place, person.

  speech normal.

Cranial nerves:

I to XII CN:intact

- Sensory system :intact
- Motor system:
- Bulk of muscle: Rt upper arm: 15cms

  Rt forearm :13.3 cms

  Rt mid thigh : 26 cms

  Rt calf muscle :20.5cms

  Lt upper arm:15.3cms

  Lt forearm :13cms

  Lt mid thigh :26cms

  Lt calf muscle :20.3cms

- Tone :normal in all four limbs.
- Deep tendon reflexes : normal.
- Plantar reflex: flexion response seen.

Musculoskeletal examination:

Power :

	Rt	Lt
Deltoid	4/5	4/5
Biceps	4/5	4/5
Triceps	4/5	4/5
Brachioradialis	4/5	4/5
Flexors of forearm	4/5	4/5
Extensors	4/5	4/5
Hand muscles	5/5	5/5
Gastrocnemius	4/5	4/5
Gluteus maximus	4/5	4/5
Gluteus minimus	4/5	4/5
Calf muscles	4/5	4/5

- GAIT: waddling gait.
- Gower sign : positive.
- All Joints, range of movements :Normal.
- Spine examination : Normal.

- Respiratory system:

  Bilateral air entry present

  Normal vesicular breath sounds heard.
- Per abdomen examination:

  soft, no organomegaly
- Cardiovascular system:

  S1,S2 Normal

  no murmur

Investigations

- Complete blood picture:

  Hb : 8.3 gm%

  Total count:6200

  N :53% L:40% E:2%

  M:5% B:0%

  Platelet count:2.2 lakh

  ESR:20

- Creatinine kinase level: 227IU/L
- LFT : Normal
- RFT : Normal
- Serum electrolytes: with in normal limits
- Nerve conduction velocity: normal
- Genetic Molecular study for dystrophin gene: report awaiting
- Muscle biopsy: not done.

SUMMARY

- 8 yr boy with progressive difficulty in walking, climbing upstairs, and frequent fall for 2yr
- O/E proximal muscle weakness, positive gower sign

Provisional diagnosis

- Progressive proximal muscular weakness
- ? Duchenne muscular dystrophy
- without cardiac or respiratory complications.