Case Presentation

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Department of Pediatrics.

- Name : xyz
- Age : 6 years
- Sex : Male
- Address : Nalgonda
- Date of admission : 17/5/2014
- Date of discharge : 2/6/2014

Chief complaints

- Fever for 2 days.
- Abnormal movements of limbs for 2 days.

History of present illness

•The child developed fever which was intermittent, moderate grade, not associated with chills and rigors, relieving with Paracetomol .

•2-3 hrs later, he developed involuntary movements of limbs which progressed gradually, resulting in difficulty in holding objects, feeding and inability to maintain posture. Movements are more noticed while child intended to do any work

➢H/o irritable behaviour is present since 1-2 months.

➢Mother noticed delay in attaining mile stones after 8 months of age.

➢No history of

•Sore throat / pyoderma

•Arthralgia

•Breathlessness/ palpitations/chest pain.

•Burning micturition.

•Abdominal pain/Vomiting/loose motions.

•Headache/Seizures/ loss of consciousness.

No history of :

- Drug intake .
- Loss of weight , appetite.
- Contact with tuberculosis.
- Head injury.
- Bladder , bowel disturbances.
- Jaundice
- Visual disturbances.
- Ear discharge .
- Regurgitation of feeds.

Antenatal history :

- Mother had regular ante-natal check-ups.
- No h/o fever with rash.
- No h/o teratogenic drug intake
- No h/o exposure to radiation.
- Mother was diagnosed with oligohydramnios in third trimester.
- No h/o diabetes mellitus/hypertension/seizures/ other complications during pregnancy.

Birth history :

- 1st by birth order.
- Product of 3rd degree consanguineous marriage.
- Full term / SGA (1.75kgs) baby born by LSCS (Indication : Oligohydramnios)
- Cried immediately after birth.
- No h/o birth asphyxia/prolonged labour.

Neonatal history :

- No h/o feeding difficulties.
- No h/o jaundice / convulsions / NICU admission/ other complications .
- On breast feeds till 7 months of age.

Developmental history :

Child was apparently normal till 8 months of age after which mother noticed delay in attaining milestones when compared to peer groups.

Gross motor milestones :

Neck holding 3 months .

Sitting with support 6 months.

Sitting without support 8 months .

Crawling and creeping 10 months .

Standing with support 3 yrs.

Walking with support 3 1/2 yrs.

Standing without support 5yrs.

Just learnt to walk with support 5 yrs 6 months.

Not able to walk without support yet. Development Quotient : 20%

- Fine motor :
- Grasps objects with both hands : 5 months.
- Palmar grasp : 7 months.
- Pincer grasp : 10 months.
- Feeding self : 16 months
- Scribbling : 2yrs DQ : 34 %

- Speech and language:
- Monosyllables : 11/2 yr
- Bisyllables : 3 yrs of age . DQ : 12%

Social mile stones :

- Social smile : 2 months
- Recognise mother : 3 months
- Play simple games : 12 months
- Follow simple commands : 15 months
- Play with others : 3yrs

DQ : 50%

- Family history :
  - The child comes from a nuclear family with one sibling.
  - Younger sister , 3yrs of age, is born by LSCS (indication : previous LSCS)
    Full term / AGA (birth weight 2.75kg)
  - H/o delayed developmental milestones present.

- Immunisation history : Immunised as per National Immunization schedule.

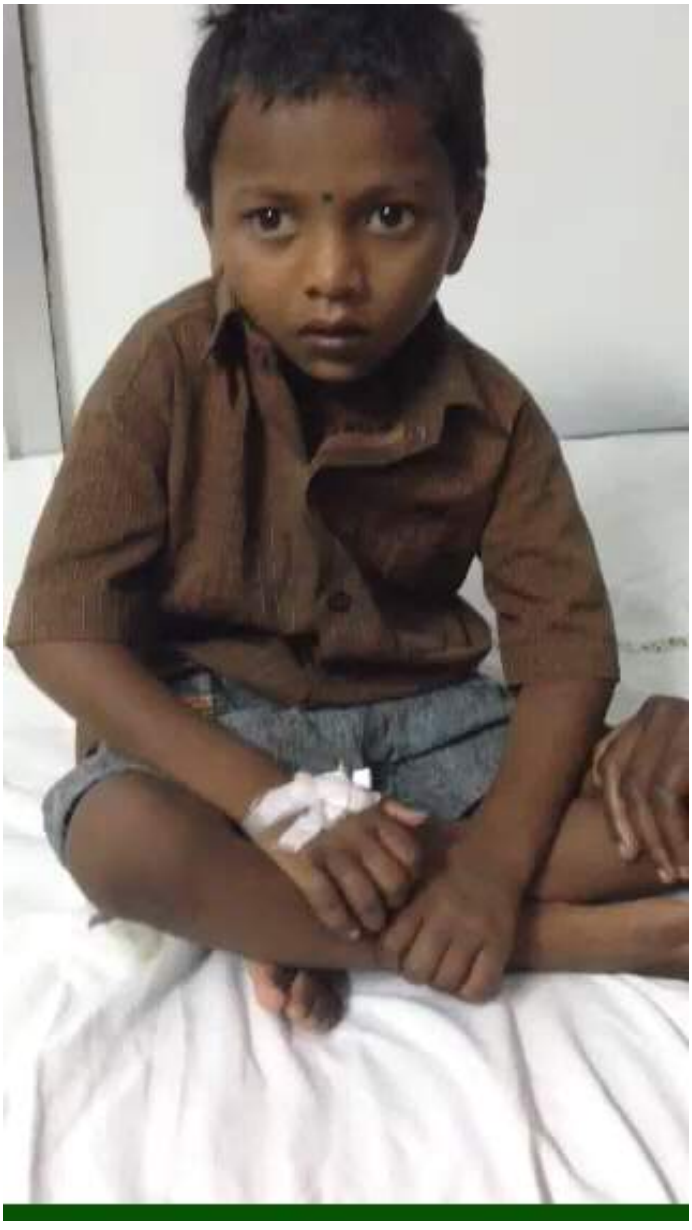
- Socio economic status : Lower middle class as per Modified Kuppuswamy scale.

•General Examination:

•Child conscious , irritable , sitting in the bed.

•Abnormal movements seen in upper limbs predominantly.

Movements are coarse, fast, repetitive and intermittent .



Head to toe examination :

- No facial dysmorphism.
- No pallor/clubbing/cyanosis/lymphadenopathy/edema/icterus.
- Chest, abdomen, lower limbs, genitalia normal.
- No neuro-cutaneous markers.

Vitals :

- Temperature : 101° F.
- Pulse rate : 110 bpm, pulse volume normal, regular rhythm, no radio-radial, radio-femoral delay. All peripheral pulses well felt.
- Respiratory rate : 30/min, abdomino-thoracic type.
- Blood pressure : 80/50 mm of mercury.

Anthropometry

	Observed
Weight(kgs)	12
Height(cms)	95
Head circumference (cms)	45

Central Nervous System :

- Higher Mental functions :
  - Conscious, irritable.
  - Intelligence : Able to understand commends, tried to perform voluntarily but was unable to perform .
  - Speech interrupted.
  - Orientation, memory could not be tested.
  - Following simple commands.
  - IQ could not be tested.

Cranial nerves

- 1st nerve : Could not be tested.
- 2nd nerve : Pupillary reflex normal. Fundus normal.
- 3rd, 4th, 6th nerves : Normal.
- 7th nerve : Normal .
- 8th, 9th, 10th,12th : Normal .
- 11th nerve : Could not be tested.

- Motor System
- Posture is unsteady with je movements more in upper limbs .
- Tremors increased on trying to catch objects.
- Bulk of muscle : normal.

Power	Right	Left
Upper Limb	3-4/5	3-4/5
Lower Limb	3-4/5	3-4/5

Tone	Right	Left
Upper limb	Increased	Increased
Lower limb	Increased	Increased

Cog wheel Rigidity present.

Reflexes :

Superficial reflexes :

- Corneal Reflex : Normal
- Conjunctival Reflex : Normal
- Abdominal Reflex : Normal
- Plantar reflex : Equivocal on both sides .

Deep Tendon Reflexes

	Right	Left
knee	4+	4+
Ankle	3+	3+
Triceps	2+	2+
Biceps	2+	2+

Sensory System :

- Touch, pain, temperature senses were intact.
- Position and vibration senses could not be tested.
- Cortical sensations could not be tested.

- Cerebellar Signs
  - Tremor : present
  - Nystagmus: absent
  - Finger nose test : could not be elicited
  - Romberg's sign : could not be elicited
  - Coordination could not be tested.
  - Gait could not be elicited.

- No primitive reflexes.

•Skull and Spine : Normal

•No signs of Meningeal irritation.

•Extra pyramidal signs :

•Rigidity present.

•Tremors present.

Provisional diagnosis

Developmental delay with mental subnormality with viral fever.

? Cerebral palsy.

? Metabolic disorder.

- CVS: S1, S2 -normal, no murmurs.

- Respiratory System: Ear, nose, throat normal.
  B/l AE equal, NVBS.

- Per Abdomen : Soft , non tender and no palpable masses.

- Musculoskeletal: Normal.

Investigations

Complete blood picture

- Hemoglobin : 13.4 gm%
- Total Leucocyte Count : 14200/cumm
- Differential leukocyte Count : N60 L34 E2 M4 B0
- Platelet Count : 2.5 lakhs
- Peripheral Smear : Normocytic Normochromic

- ESR : 20 mm
- CRP : Negative
- CUE : Normal
- CXR : Normal Study

INVESTIGATIONS

Liver function tests

- TSB : 0.6mg/dl
- DB : 0.17 mg/dl
- AST : 20 IU/L
- ALT : 11 IU/L
- ALP : 220 IU/L
- T.Proteins : 7.7 gm/dl
- S. albumin : 4.3 gm/dl
- A/G ratio : 1.2

Renal function tests

- B. Urea 28 mg/dl
- S. Creatinine 0.5 mg/dl

Serum Electrolytes

- S. Calcium 8.7mg/dl
- S. Sodium 146 mmol/l
- S. Potassium 4 mmol/l
- S. chloride 104 mmol/l

Treatment

- Child was admitted and given symptomatic treatment with

1. Paracetomol 15mg/kg/dose
2. Cefotaxime 100mg/kg/day

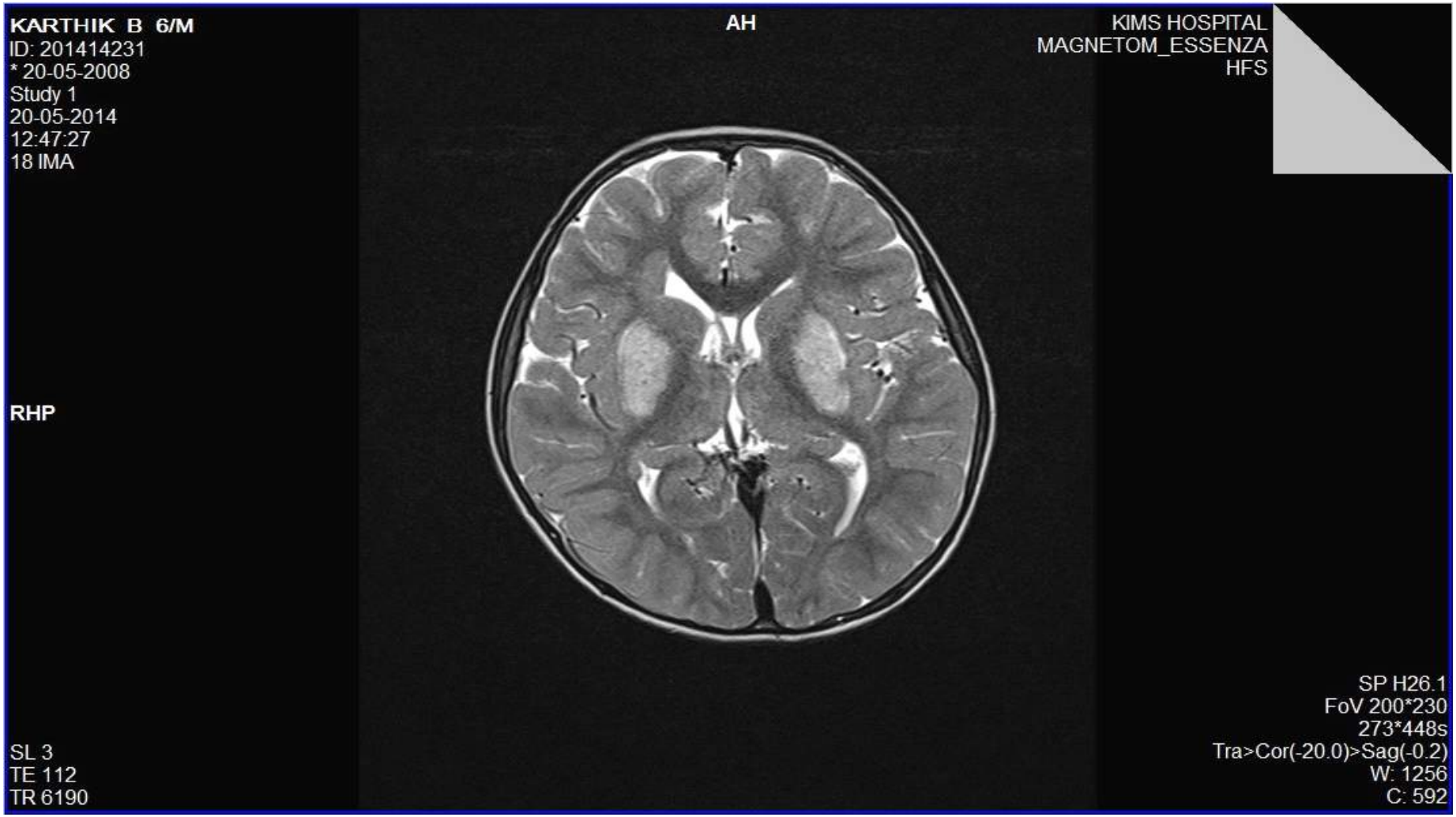
- Fever subsided by 3rd day of admission but involuntary movements persisted.

- MRI Brain was planned in view of mental subnormality, developmental delay, abnormal movements.

Radio-imaging

MRI Brain:

- T2W/T2 hyperintensity across the bilateral putamen, right caudate tail, bilateral ventricle s/o Wilson disease
- Right sided peri ventricular band heterotopia.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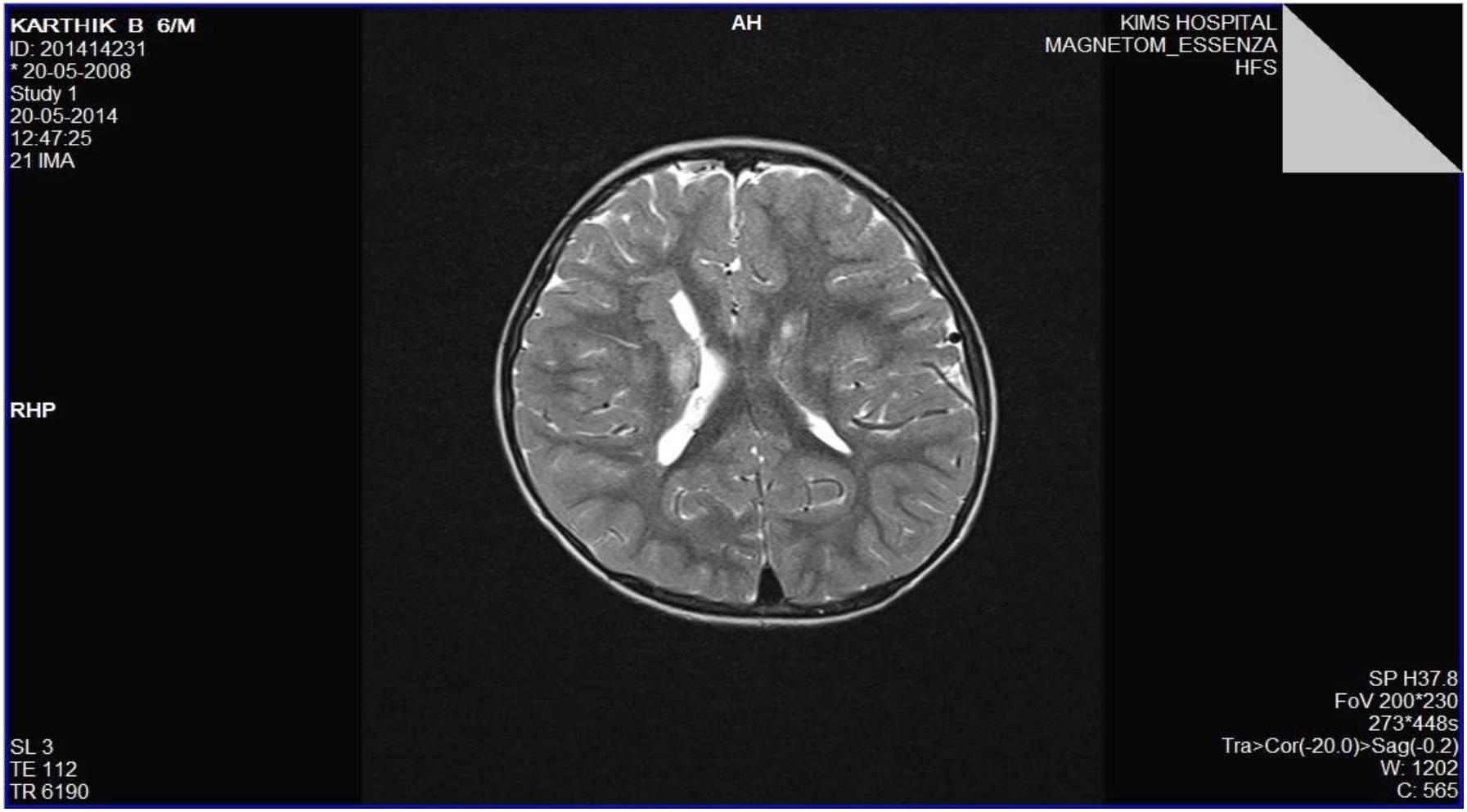
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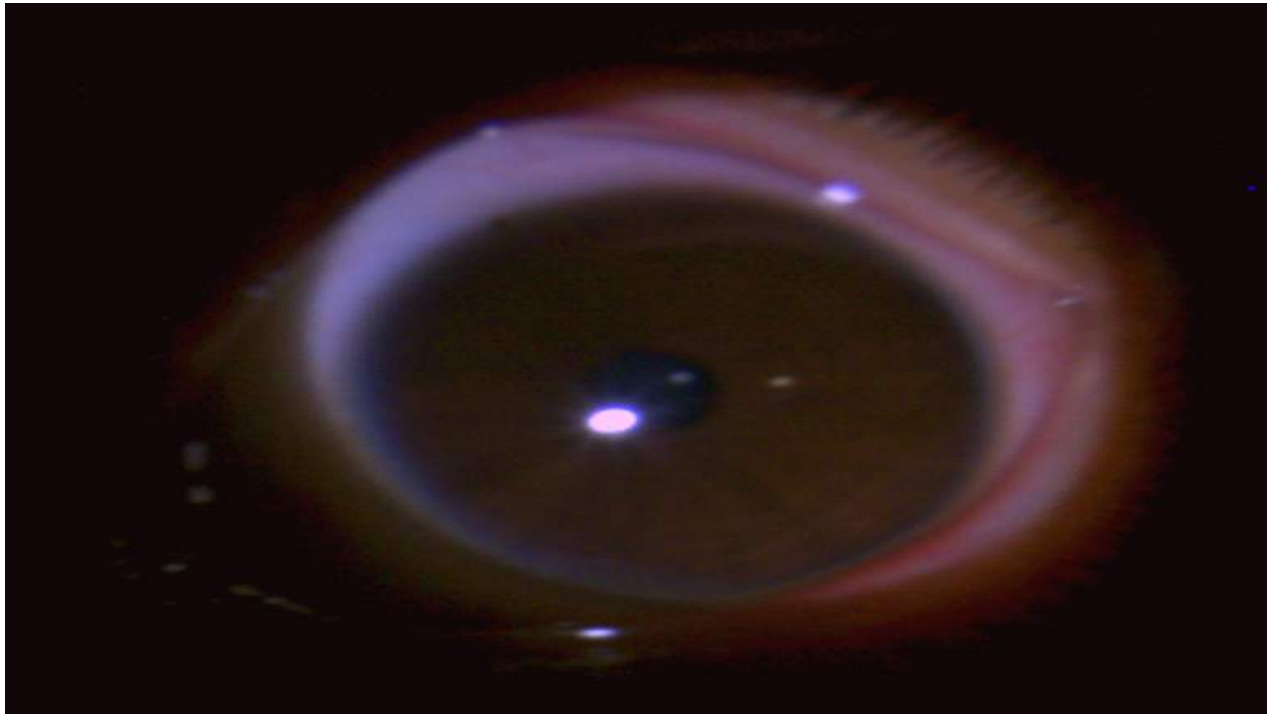
USG Abdomen :

- Liver normal size, mildly echogenic.
- 5-6 mm calculi found in mid and lower pole of right kidney and upper pole of left kidney.

Opinion of ophthalmologist

•Slit lamp examination revealed KF rings more in left eye compared to right eye.



Provisional diagnosis

- Wilson disease

Investigations

- S. Ceruloplasmin 10 mg/dl ↓ (19 – 57 mg/dl)
- S. Copper 120 µ gm/dl (70 – 140 micro gm/dl)
- 24 hrs Urinary Copper 5 µ gms/day (15 to 70 micro grams /day)

- Pencillamine was started at 20 mg/kg/day , 2 doses were given and urinary copper sent again.

- 24 hr urinary copper ↑ 110 µ gms / day.

Final Diagnosis

- Wilson disease.

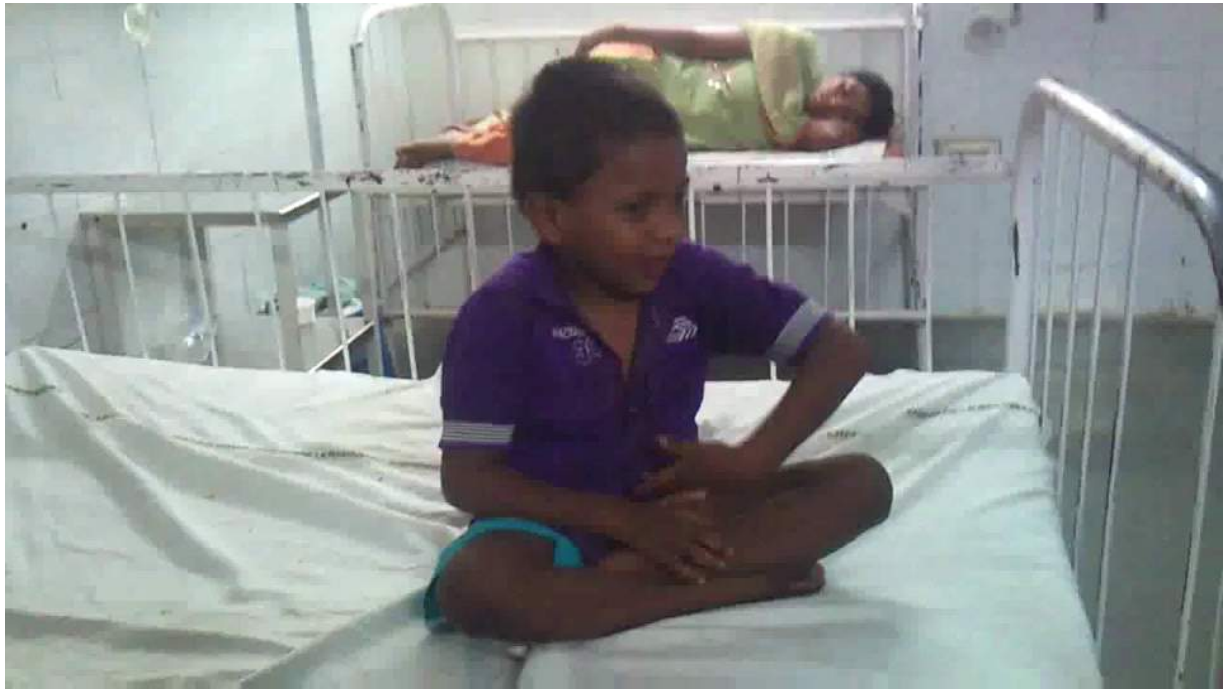
Treatment

- Tab Pencillamine 20mg/kg/day in 2 divided doses
- Tab Pyridoxine 3 mg /kg/day

- Other sibling was also screened . Her investigations were normal.
- Zinc supplements were given to the sibling and was advised for regular follow up.

Follow up :

- The child came for follow up after 1 month.
- Intentional tremors have decreased.
- Able to perform daily activities like eating etc.
- Able to walk few steps without support.



THANK-YOU