KLIPPEL FEIL SYNDROME

– A CASE REPORT

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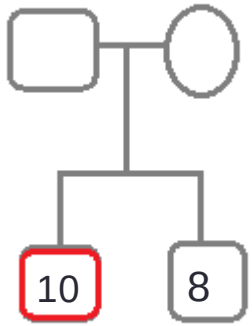
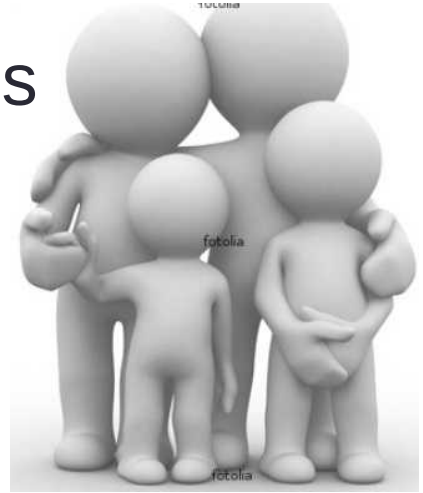
CASE HISTORY

11year old male child 1st born of non consanguineous marriage came with C/O

- Shoulder deformity since birth
- Severe pain in neck, both upper limbs for past 2 months

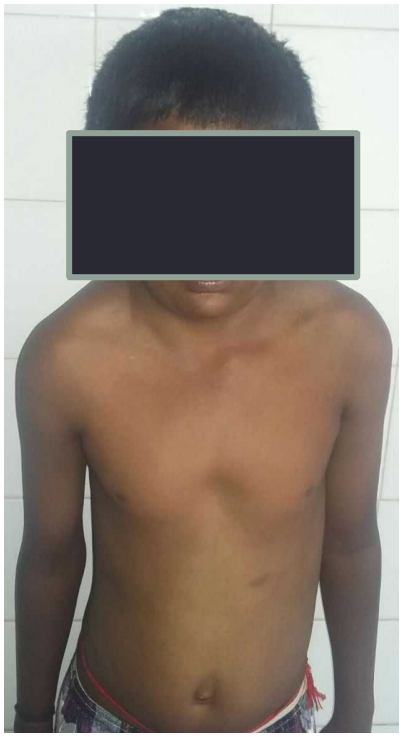
- Normal Developmental history
- Good scholastic performance
- No history of contact with open case of TB
- No family history of similar illness



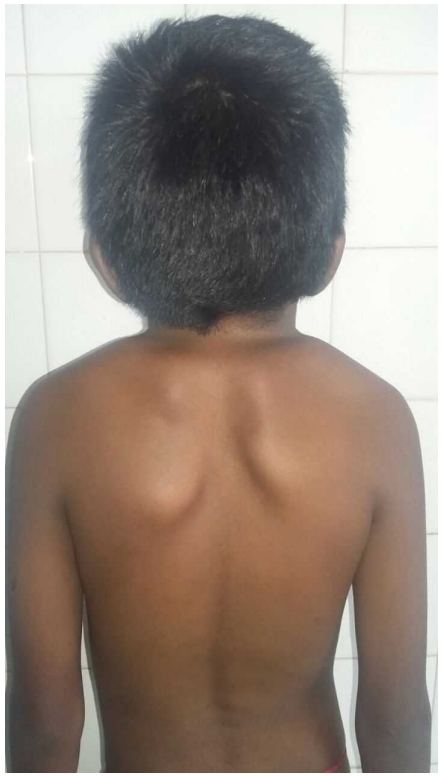


O/E:

- Short neck
- Low hair line



- Elevation of scapula (Sprengel shoulder)
- Scoliosis



- General examination – normal

- Systemic Examination

- CVS
- RS
- P/A

Normal

- CNS

Alert

Cranial nerve examination – normal

- Fundus examination – normal;no papilledema

Motor system examination:

- Musculoskeletal system-child not able to abduct and elevate the shoulder
- Able to move head sidewards
- Flexion/ extension of head restricted
- All other joint movements normal

- Spine – Scoliosis
- Cranium – normal
- No cerebellar signs

Sensory system examination: normal

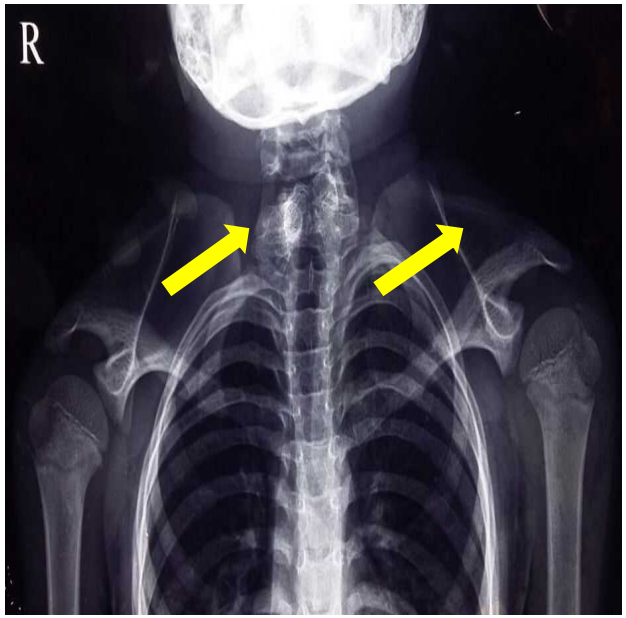
INVESTIGATIONS

- Routine blood investigations were normal
- USG abdomen – B/L kidneys normal.
  No significant abnormalities detected
- ECHO – normal study
- ENT Evaluation -No Conductive or Sensory neural hearing loss

- X-RAY cervical spine

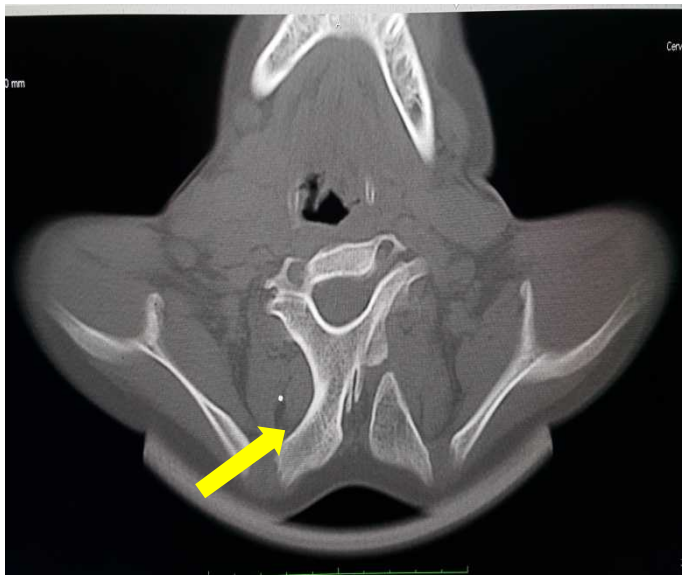
  Fusion of C5 – C7 vertebra

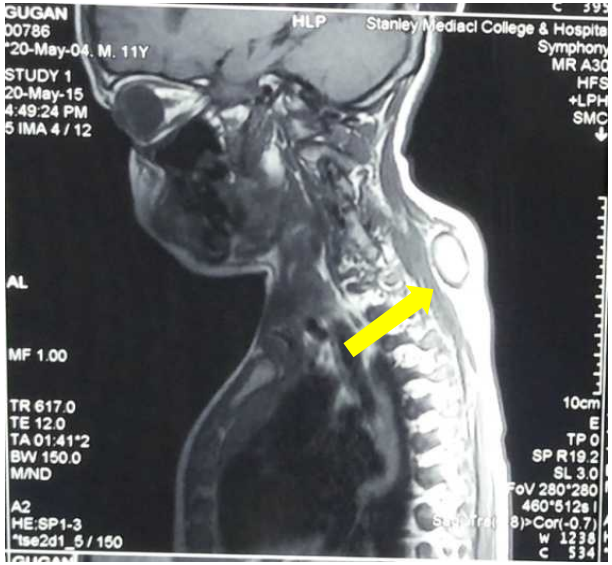
  High riding scapula



- CT Head & Neck :

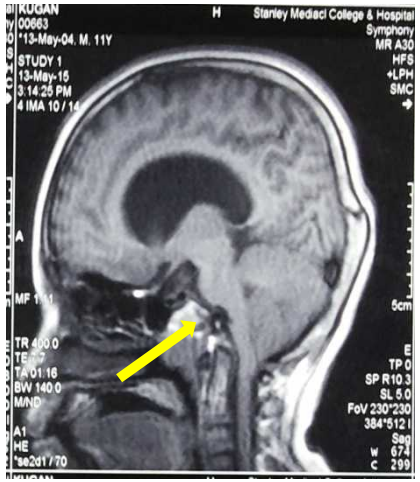
Bilateral omovertebral bone (abnormal bone that extends from the medial border of scapula to the spinous process, lamina or transverse process of C5, C6 & C7



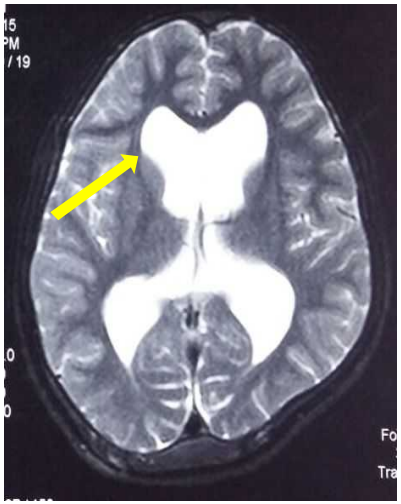


- MRI Brain & Spine :

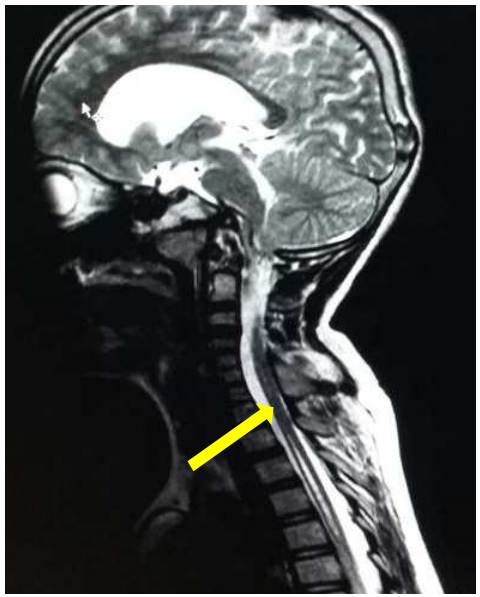
Aqueductal stenosis & Arnold Chiari malformation TypeII



Obstructive hydrocephalus



Syringomyelia C3- L3



DIAGNOSIS

- KLIPPEL FEIL SYNDROME

TREATMENT:

- Sub occipital craniectomy
- Foramen magnum decompression

} for Chiari malformation

- C5,6,7 hemilaminectomy - for cervical fusion of vertebra
- Hydrocephalus – VP shunt
- Physiotherapy
- Postoperatively pain subsided significantly

DISCUSSION

HISTORY OF KLIPPEL FEIL:

- This syndrome has been described in a mummy from 500 B.C.
- Date back from writings – 13 to 16th century
- First described by Maurice Klippel & Andrew Feil in 1912
- Klippel and Feil pointed this disorder in a French tailor with a short neck
- Also called as Brevicollis or congenital synostosis of cervical vertebra.

CLINICAL FEATURES

- Klippel Feil Syndrome(KFS) is congenital fusion of cervical vertebrae, whether it involves 2 segments, congenital block vertebra, or the entire cervical spine
- Prevalence 1 in 40000 live births

- Classical triad:
  - ✓ Low hair line
  - ✓ Short neck
  - ✓ Restricted cervical motion
- Fewer than 50% have this triad. Our patient has all the 3 triad.

Class of Klippel-Feil syndrome	Inheritance	Vertebral fusion and associated anomalies
KF 1	Autosomal recessive	Rostral fusion at C1 and severe associated anomalies (short neck, cardiac defects, and craniofacial anomalies)
KF 2	Autosomal dominant	C2-3 fusion And possible craniofacial anomalies,Syringomyelia
KF 3	Reduced penetrance	Singular isolated fusion, most rostral at C3
KF 4	X-linked inheritance	Vertebral fusion and ocular anomalies

PATHOGENESIS

- Failure of normal segmentation of cervical somites at 3-8 weeks gestation- multiple fused cervical segments.
- SGM 1 gene is disrupted by a heritable paracentric inversion of chromosome 8.

Common associated anomalies:

- Scoliosis (60%) - Congenital or compensatory
- Renal anomalies (35%) – U/L agenesis, double collecting system, renal ectopia, horseshoe kidney, hydronephrosis
- Sprengel deformity (30%)
- Deafness &mixed (30%) – conductive, sensorineural
- Synkinesis / Mirror motions (20%)
- Congenital heart disease (14%) - VSD and PDA

Less common :

- Ptosis
- Facial nerve palsy
- Syndactyly
- Hypoplastic thumb
- Upper extremity hypoplasia
- Neurenteric cyst
- Syringomyelia (<1%)
- Arnold chiari malformations (1-2%)
- Aqueductal stenosis (0.5 – 1%)
- Hydrocephalus (<1%)

TREATMENT

- Treatment is symptomatic.
- Traction, cervical collar, analgesics
- Surgical intervention
- For cosmetic problems- corrective osteotomy, soft tissue procedures, Zplasty, muscle resection

CONCLUSION

- This Klippel Feil syndrome is not so rare. But it has presented with a rarer symptom and we present this case because of its rarest associated anomalies

AWARENESS !!


TODAY!
Klippel-Feil Syndrome
Awareness Day
August 6th
#KFStroll
Klippel-Feil Syndrome Freedom

REFERENCES

- *1.Klippel-Feil syndrome: a case report. Kawu Ahidjo A, Salami Olayinka AO, Ayokunle O.Niger Postgrad Med J. 2010 Dec;17(4):320-3.PMID: 21809613*
- *2. Sprengel's deformity in Klippel-Feil syndrome. Samartzis D, Herman J, Lubicky JP, Shen FH.Spine (Phila Pa 1976). 2007 Aug 15;32(18):E512-6.*
- *3. Klippel-Feil syndrome associated with congenital cervical dislocation: report of an autopsy case. Shintaku M, Wada K, Koyama T, Kohno H, Sakamoto T, Hida S.Clin Neuropathol. 2013 Jan-Feb;32(1):51-7. doi:10.5414/NP300498.*

THANK YOU

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