International Journal of Science and Research (IJSR)
ISSN: 2319-7064
Impact Factor 2024: 7.101

Klippel-Feil Syndrome: A Case Study of Congenital Cervical Fusion and Associated Anomalies in a Newborn

Dr. Poonam Thakur[1], Dr. Pritesh. A. Pawar[2], Dr. Shivshakti Pawar[3]

[1]Assistant Professor, Department of Pediatrics, VM GMC, Solapur

[2]Consultant Cardiologist Yashodhara Superspeciality Hospital, Solapur

[3]District Survillence Officer, District Health Office, Nanded

Abstract: ***In this case study, we explore a rare instance of Klippel-Feil syndrome (KFS), a congenital disorder marked by the fusion of cervical vertebrae, as observed in a male newborn admitted to the neonatal intensive care unit (NICU) at Basaweshwar Hospital. Born via cesarean section to a 23-year-old mother with no antenatal care, the infant presented with a short neck, low hairline, restricted neck mobility, and additional anomalies like encephalocele, Sprengel's deformity, and respiratory distress. Diagnostic imaging, including X-rays and MRI, confirmed the fusion of C2 to C5 vertebrae alongside communicating hydrocephalus. In my view, this case underscores the critical need for early diagnosis and intervention, particularly in unmonitored pregnancies where such conditions might otherwise go undetected until birth. It is evident that KFS, though rare with an incidence of 1 in 40,000–42,000 newborns, carries a complex array of associated anomalies that challenge neonatal management. This report not only highlights the genetic underpinnings mutations in GDF6 and GDF3 genes but also reflects on the symptomatic treatment approaches, offering insights into the prognosis when addressed promptly. Taking this further, the case invites discussion on the broader implications for prenatal screening and the management of cranio-cervical instability in such patients.***

Keywords: Klippel-Feil syndrome, congenital cervical fusion, encephalocele, neonatal respiratory distress, Sprengel's deformity

1. Case Report

A male baby weighing 2.5kg, was born to 23yrs old primi mother through cessarean- section in our hospital i.e. Basaweshwar hospital mother was an unbooked case with no antenatal check ups, there is no history consanginuous marriage. The baby was admitted to N.I.C.U. for respiratory distress, shortness of neck, very low hairline & ability of movement of neck limited, the baby had swelling of neck (encephalocele) congenital elevation of scapula (sprengel's deformity) & sacral dimple.

On Examination:

Anterior fontanelle: 3x2.5cms

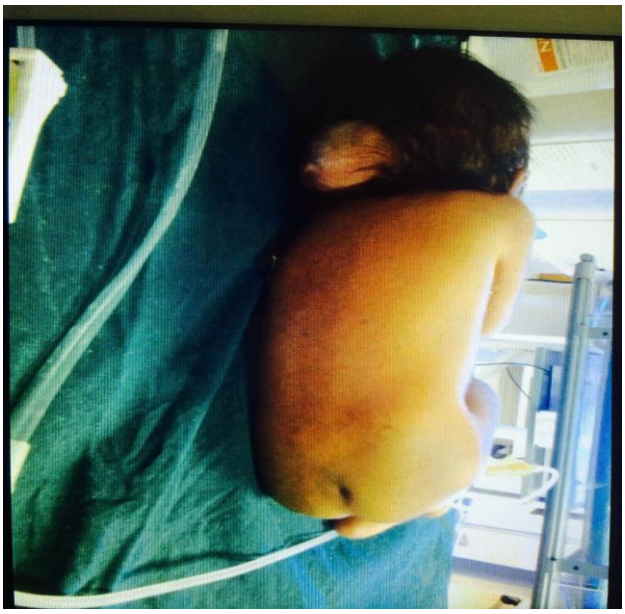
Respiratory system: Respiratory rate:80/min, subcostal, intercostals retractions present, nasal flare present, no cyanosis, no grunt (downe's score:4/10)

Cardiovascular system: S_1, S_2 heard, no murmur

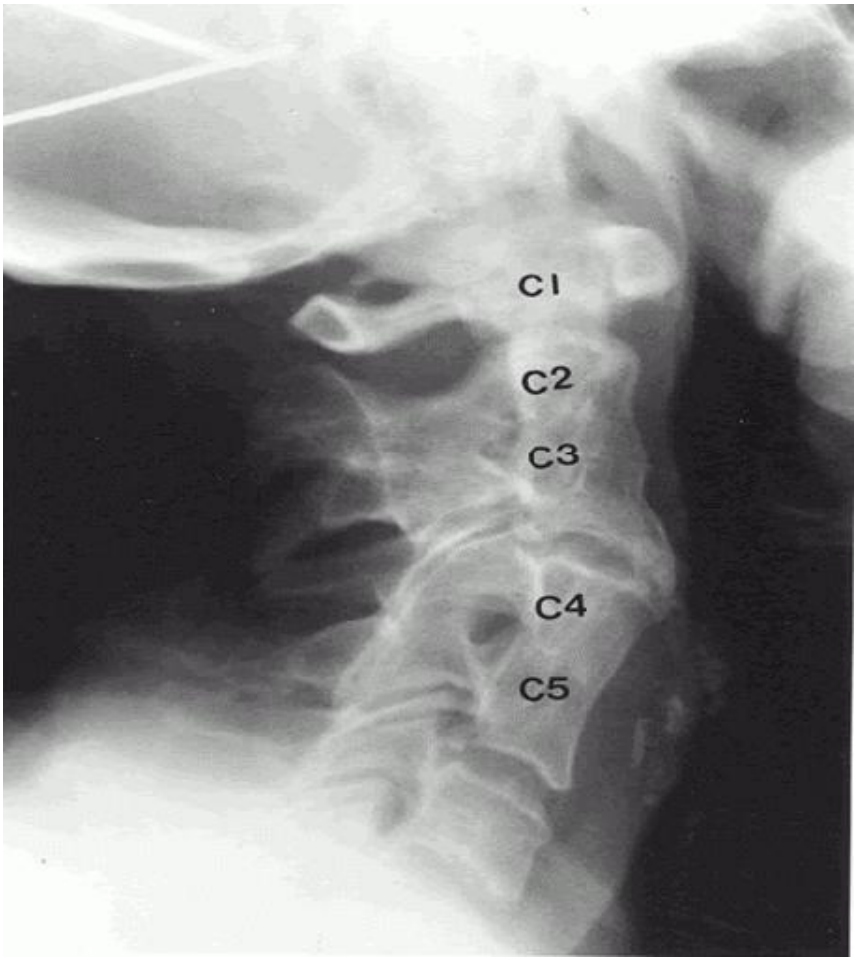
Per abdomen: soft,no organomegaly

Central nervous system: cry, activity was fair.

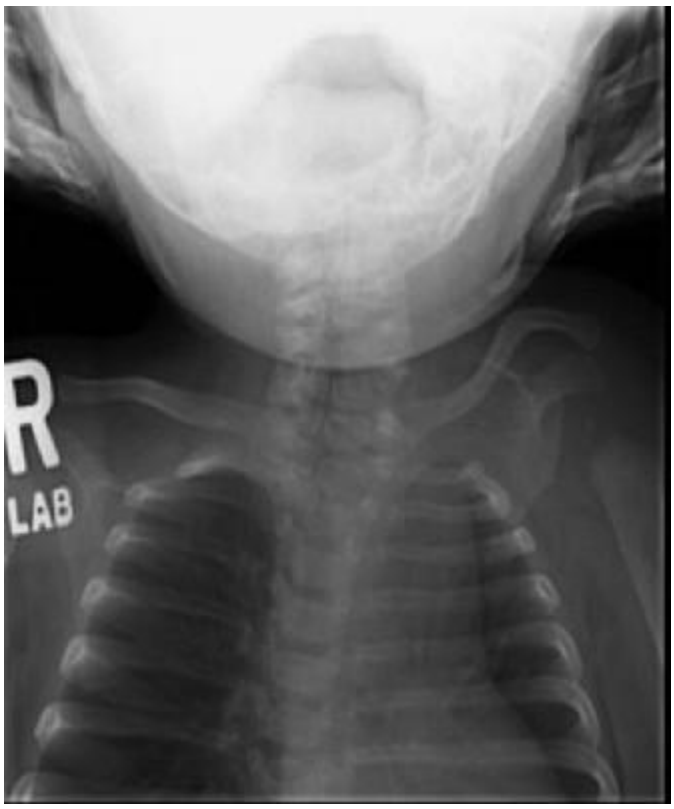
Routine blood investigations were normal, CRP was negative



BABY shows swelling of neck (encephalocele), shortness of neck, very low hairline.

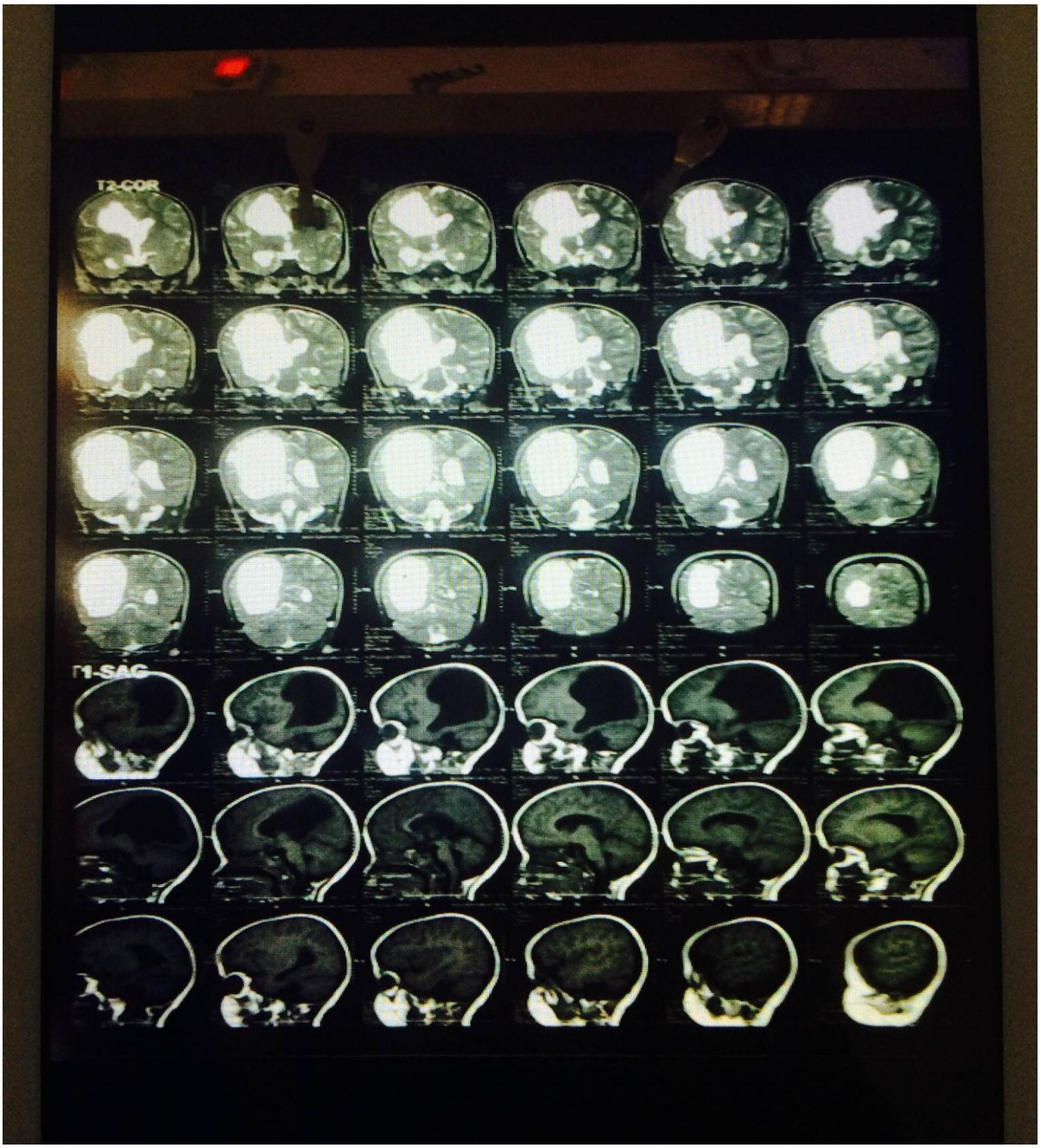


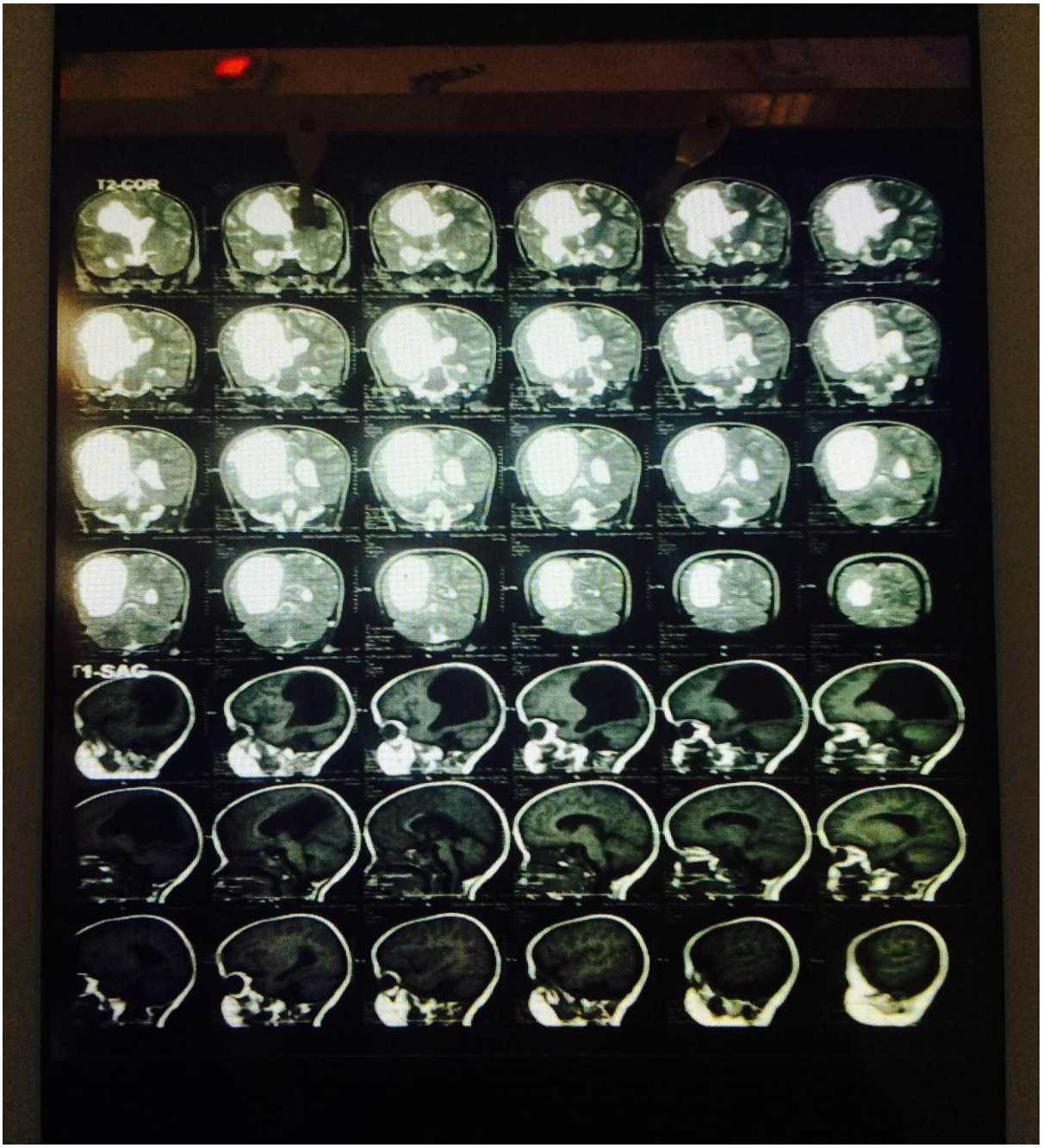
Lateral X-Ray Neck: FUSION OF C2, C3, C4 & C5 VERTEBRAE

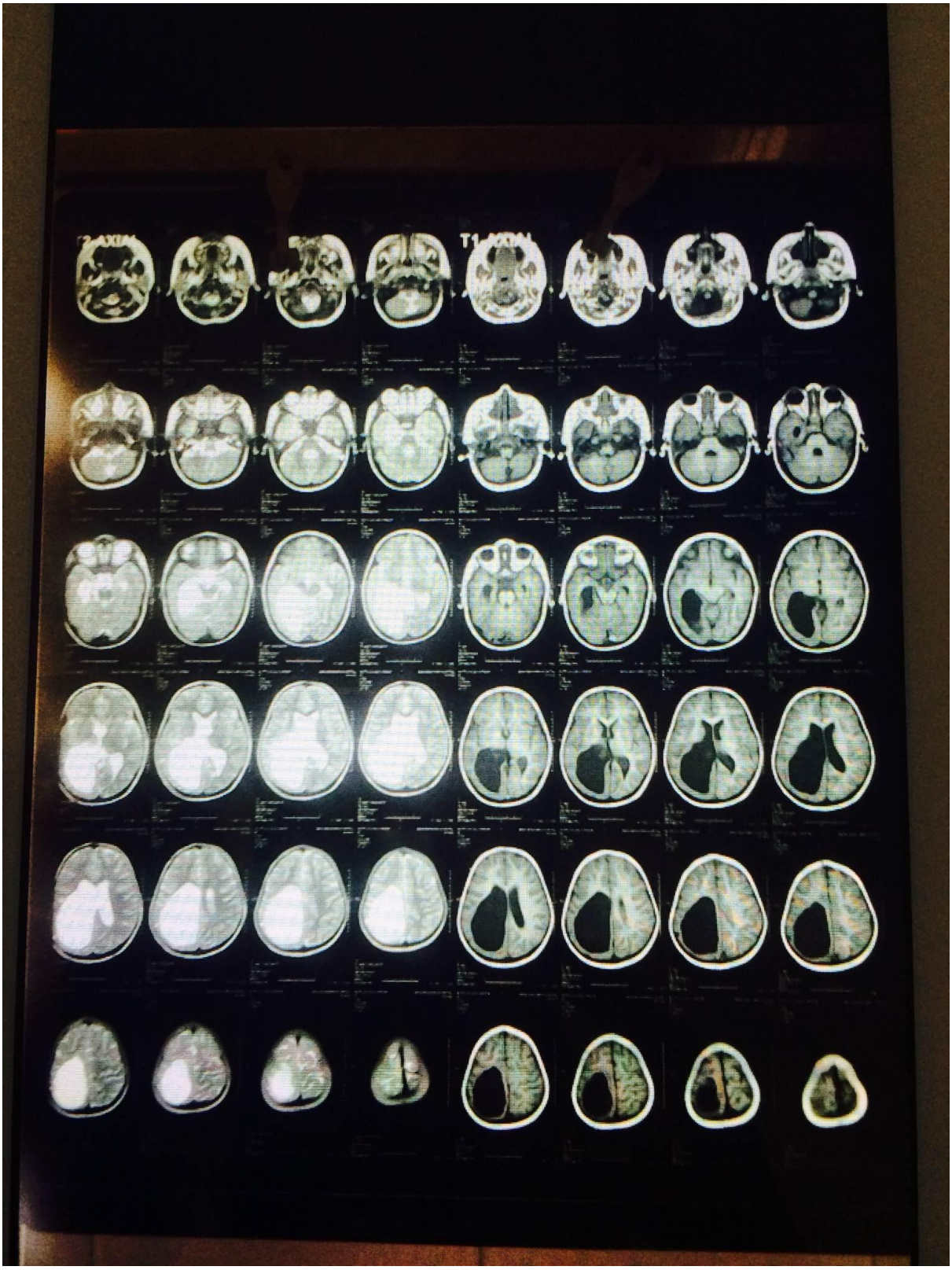


X-Ray AP Neck & Chest: FUSION OF C2, C3, C4 & C5 VERTEBRAE.

MRI BRAIN: Communicating hydrocephalus with encephalocele.




T2-COR
T1-SAG



2. Discussion

Klippel feil syndrome is a genetic disorder caused due to mutation in the GDF6 and GDF3 genes have also been identified to cause the disease. These mutations can be inherited in 2 ways autosomal dominant inheritance and autosomal recessive inheritance, it is classified into 3 types,1) Fusion of C2 and C3 with occipitalization of atlas 2) Long fusion below C2 with an abnormal occipital-cervical junction.3) A single open interspace between two fused segments. Cervical spine motion is concentrated at single open articulation. Associated anomalies are scoliosis, spins bifida, anomalies of kidney and ribs, cleft palate, respiratory problems, heart malformation, short stature, encephalocele, winging of scapula (sprengel's deformity). Neonates present at birth with short neck, low hairline, limited ability to move the neck and respiratory distress. Diagnosis is confirmed by taking lateral neck x-ray which shows congenital fusion of any 2 cervical vertebrae, M.R.I brain to see for hydrocephalus or any other cranial

anomalies, treatment of klippel feil syndrome is symptomatic to relieve cervical and craniocervical instability and constriction of spinal cord to rcorrect scoliosis, adjacent segment disease and scoliosis are 2 examples of common symptoms associated with klippel feil syndrome. They can be corrected with cervical disc arthroplasty and alternate is total disc replacement. Prognosis for most individuals with Klippel Feil Syndrome is good if the disorder is treated early on and appropriately.

References

[1] Klippel M, Feil A (1912). "Un cas d'absence des vertebres cervicales. Avec cage thoracique remontant jusqu'a la base du crane (cage thoracique cervicale)". *Nouv Iconog Salpetriere* **25**: 223–250.
[2] Andrews, James, William; Berger, Timothy; Elston, Dirk (2005). *Andrews' Diseases of the Skin: Clinical Dermatology*. (10th ed.). Saunders. ISBN 0-7216-2921-0.
[3] "Klippel-Feil Syndrome". HONselect.
[4] Farsetti P, Weinstein SL, Caterini R, De Maio F, Ippolito E (May 2003). "Sprengel's deformity: long-term follow-up study of 22 cases". *J Pediatr Orthop B* **12** (3): 202–10. doi:10.1097/01.bpb.0000049568.52224.1e. PMID 12703036. Sarwark, JF; LaBella, CR, (2010). *Pediatric Orthopaedics and Sports Injuries: A Quick Reference Guide*. Elk Grove Village IL: American Academy of Pediatrics. pp. 231–4.