Print ISSN: 2321-6379
Online ISSN: 2321-595X

Case Report

A Rare Case of VACTERL Association

Jeff Walter Rajadurai[1], S Mohan[2], A Diana[3], B Shindya[3], S Priyanka[3]

[1]Orthopaedics and Joint Replacement Surgeon, Subam Clinic, Coimbatore, Tamil Nadu, India, [2]Principal, Department of Pharmaceutical Bio Technology, Karpagam College of Pharmacy, Coimbatore, Tamil Nadu, India, [3]Pharm D, Department of Pharmacy Practice, Karpagam College of Pharmacy, Coimbatore, Tamil Nadu, India

Abstract

Klippel-Feil syndrome (KFS) is a rare skeletal disorder primarily characterized by abnormal union or fusion of two or more cervical vertebrae. It also includes multiple system anomalies. Herewith, we report a case of 22 years old male who presented with multiple skeletal anomalies. On subsequent radiological investigations, patient was diagnosed to have Klippel-Feil syndrome with type IV club hand.

Key words: Cervical vertebra fusion, Club hand, Klippel-Feil syndrome

INTRODUCTION

Klippel-Feil Syndrome was originally described in 1912 by doctors Maurice Klippel and Andre Feil from France.[1,2] They described patients who had a short and webbed neck; decreased range of motion (ROM) in the cervical spine. These patients had a low hairline. Reports estimate that the condition occurs in approximately 1 in 40,000-42,000 live births and it affects females more frequently than males.[2]

Aim

This study aims to present a rare case report on VACTERL association with orthopedic manifestation.

CASE REPORT

A 22-year-old male patient presented with deformity of both hand and forearm since birth and swelling over the left knee for the past 1 week

H/O pain – insidious in onset, pricking in nature, intermittent, radiating to left leg, aggravated on doing work, relieved at rest

Access this article online

www.ijss-sn.com

Month of Submission : 04-2020
Month of Peer Review : 05-2020
Month of Acceptance : 05-2020
Month of Publishing : 06-2020

H/O fever – low grade, no chills and rigors, no evening rise of temperature

No H/O loss of weight and appetite

He was the second child born to parents of Grade IV consanguinity

Natal history: Full term, forceps delivery, H/O delayed milestones, vaccinated up to date.

Past history: H/O bleeding per rectum on and off for past 5years and was diagnosed to havehemorrhoids and under irregular treatment.No H/O Diabetes,systemic hypertension bronchial asthma.

Prenatal history: He was the second child born to parents of Grade IV consanguinity.Mother conceived spontaneously, H/O drug consumption to terminate pregnancy by 3rd month of pregnancy.No H/O fever, diabetes mellitus, systemic hypertension during the course of pregnancy.

Personal history: He takes a mixed diet. Sleep is adequate. He is not a smoker and not an alcoholic.

Family history: Siblings and other family members do not have any congenital deformities.

Treatment History: Patient never had any treatment for the congenital deformity of the hand and forehand.

General Physical Examination: He is consious, cooperative, well oriented to time,place and person.

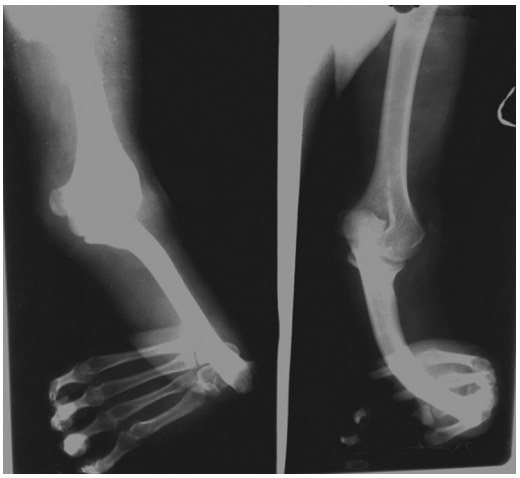
Corresponding Author: Dr. Jeff Walter Rajadurai, Orthopaedics and Joint Replacement Surgeon, Subam Clinic, Coimbatore, Tamil Nadu, India.

Not pale, not icteric, not cyanosed, no clubbing, no edema, no significant lymphadenopathy, no neuro-cutaneous markers.

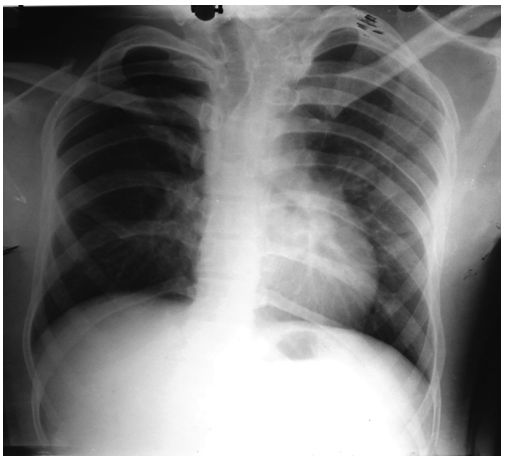
On Examination

Vitals: Stable

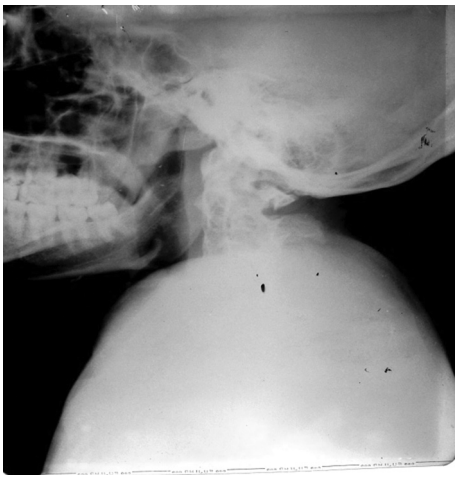
a. Radiograph right forearm with hand – AP and lateral view: Complete absence of radius with malrotated and superior angulated wrist with complete absence of thumb; fusion of the 2nd and 3rd MC and phalanx on the right hand with thinned out 2nd MC – Type IV club or mitten hand



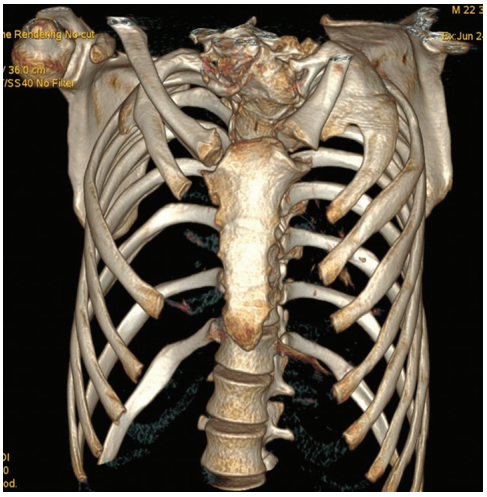
b. Radiograph left forearm with hand – AP and lateral view: Complete absence of radius with malrotated and superior angulated wrist with complete absence of thumb – Type IV club hand



c. Frontal chest radiograph – mild scoliosis of upper vertebra, widening of right intercostal space; left scapula higher in position when compared to right; bilateral 1st and 2nd ribs fused



d. Radiograph of cervical spine – lateral shows multiple cervical vertebra fusion with straightening of cervical spine with loss of cervical lordosis and short neck



e. 3D volume rendered image shows elevated left scapula – Sprengel deformity

f. 3D volume rendered image: Fusion of left 1st and 2nd ribs.

Other Investigations

- Blood investigations CRP – positive, rheumatoid factor – negative. Other blood reports under normal limits
- CT thorax – no significant abnormalities
- USG abdomen and pelvis – left pelvis ectopic kidney (left kidney is not visualized in the lumbar region and seen in the left hemipelvis posterior to the bladder assuring 9.8 × 3.9 cm)
- ECHO cardiogram: Mild MR and mild AR.

Management

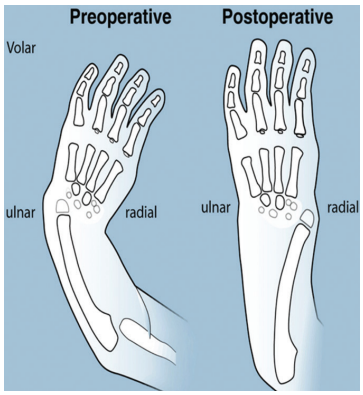
- Non-operative
  - Infants – splints are difficult to apply
  - Wrist and elbow physiotherapy
  - Serial casts to stretch tight structure.

- Operative
  - Centralization

  Indications:
  - Good elbow motion and biceps function
  - 6–12 months baby
  - Capable of tendon transfer.

- Radialization
  - Conversion of ulna into radius
  - Hand and carpal bones are translocated to ulnar side
  - Tendon repair
  - Low recurrence.

As our patient was adapted towards his various deformities and was symptom free, he was well managed conservatively.



Syndromic Association

HOLT ORAM (chromosome 12)	Thrombocytopenia – absent radius syndrome
Klippel–Feil syndrome	Fanconi anemia
VACTERL	
V – Vertebral defects	
A – Anal atresia	
C – Cardiac abnormalities	
T – Tracheoesophageal fistula	
E – Esophageal atresia	
R – Renal defects	
L – Limb anomalies	

CONCLUSION

- A high degree of suspicion and knowledge of various combinations of congenital anomalies must

be in kept in mind, in diagnosing of such complex cases

- These are rare congenital anomalies, which need team management
- Parental counseling also forms an integral part of this management.

REFERENCE

1. Nguyen VD, Tyrrel R. Klippel-Feil syndrome: Patterns of bony fusion and wasp-waist sign. Skeletal Radiol 1993;22:519-23.
2. Thomsen MN, Schneider U, Weber M, Johannisson R, Niethard FU. Scoliosis and congenital anomalies associated with Klippel-Feil syndrome Types I-III. Spine (Phila Pa 1976) 1997;22:396-401.

How to cite this article: Rajadurai JW, Mohan S, Diana A, Shindya B. A Rare Case of VACTERL Association. Int J Sci Stud 2020;8(3):8-11.

Source of Support: Nil, **Conflicts of Interest:** None declared.