AN INTERESTING CASE OF CHRONIC KIDNEY DISEASE WITH CONGENITAL SKELETAL ABNORMALITY

GOWRI SHANKAR

Department of General Medicine,
STANLEY MEDICAL COLLEGE AND HOSPITAL

Abstract : A young male presented with breathlessness. The patient was investigated and diagnosed to have Chronic Kidney Disease and Type 2 Klippel Fiel Syndrome. He improved after haemodialysis and is on regular follow up.

Keyword : Chronic kidney disease, Klippel Fiel Syndrome, Sprengel shoulder haemodialysis, Omovertebral bone, renal agenesis.

21 years old male was admitted with the complaints of breathlessness and easy fatigability of 6 months duration. His breathlessness was gradually progressive and he was in class ii dyspnoea at the time of admission. It was not associated with chest pain, palpitation, profuse sweating, orthopnoea or paroxysmal nocturnal dyspnoea. There was also no history of cough with expectoration, evening rise of temperature or haemoptysis. Patient had easy fatigueability. There was no history of hematemesis or melena or any other bleeding diathesis. There was reduced urine output without dysuria, urgency or dribbling of urine. There was no history of hypertension, diabetes, coronary artery disease or tuberculosis. Personal history and family history were not significant.

General examination: conscious, oriented, afebrile, pale, not icteric, not cyanosed and no clubbing. There was no significant generalised lymphadenopathy or pedal edema. There was low hair line with absent anti helix in left pinna. He had short neck with Height measuring 172 cm, Neck measuring 12 cm, Ratio: 14.3:1. Scoliosis present with convexity to right. Movements of the neck were restricted and he had left Sprengels shoulder. Fundus examination- Normal.

Vitals: pulse- 98/min, RR- 38/min, BP- 170/76 mmHg. CVS,RS,ABDOMEN & CNS examination were normal.

Investigations: CBC: Hb-8 gm, Urea-158 mg, Creatinine-7.4 mg, Na-142.7 Meq, K-5.13 Meq, Cl-94 Meq . Sr. Calcium 8.4mg%. RBS:- 107mg%, Urine routine examination:- Albumin 1 +, Sugar-nil, Deposits- 5-6 pus cells, 1-2 epithelial cells. CXR –Left omovertebral bone. ECG – NSR, WNL. ECHO-NORMAL STUDY. USG ABDOMEN: Left ectopic kidney. Right kidney not visualised. CT ABDOMEN: - Right kidney agenesis with left malrotated contracted ectopic pelvic kidney. HIV, HBsAG, ANTI HCV were negative. His autoimmune workup showed ANA and RA-negative. C3 levels were normal. ENT opinion was sought which revealed mild conductive hearing loss on left side. Nephrologist opinion was obtained. Patient underwent haemodialysis.
CT & MRI NECK:-Congenital block vertebra at C5/C6 level. C2,C3 right sided facets are fused. The left scapula is elevated, rotated with shortening of its vertebral border and a large omovertebral bone uniting with the spinous process of the C5/C6 vertebral body. No e/o syrinx or spina bifida. CT CHEST:-Scoliosis with normal lung parenchyma.

ECTOPIC LEFT KIDNEY

MRI ABDOMEN:-Both kidneys are absent in the renal fossa. Malrotated ectopic left kidney noted just superior to the dome of urinary bladder. The left ureter is tortuous and prominent and inserts on the left side of bladder. No evidence of hydronephrosis. Right kidney-Agenesis.

FINAL DIAGNOSIS:-Type II KLIPPEL FEIL ANOMALY with LEFT SPRENGEL'S SHOULDER.RIGHT RENAL AGENESIS with CONTRACTED SOLITARY LEFT PELVIC KIDNEY.

MANAGEMENT:-Our patient is on maintenance hemodialysis. He is on regular follow up.

CASE DISCUSSION: Our case is presented because of the rare associations of skeletal anomalies with chronic kidney disease. The cause of renal failure could not be ascertained as he presented in an advanced stage requiring haemodialysis. Klippel–Feil syndrome - Maurice Klippel and André Feil from France described this syndrome in 1912. It is characterized by the congenital fusion of any 2 of the 7 cervical vertebrae. Classic clinical triad with low posterior hairline, short neck and limitation of neck movements. ETIOLOGY:-Unknown. Autosomal dominant inheritance is especially associated with C2-C3 fusion, with gene mapped on locus 8q22.2 (mutations in GDF6). Autosomal recessive inheritance is especially associated with C5-C6 fusion. Incidence 1 in 42,000, female predominance.

FEIL'S CLASSIFICATION:-
Type I - massive fusion of many cervical and upper thoracic vertebrae with synostosis. Type II - fusion of only 1 or 2 vertebrae (with hemi vertebrae, scoliosis, occipito-atlantoid fusion). Type III - presence of lower thoracic and upper lumbar spine anomalies with I/II. Type IV - sacral agenesis

SAMARTZIS and colleagues suggested a new classification system.

Type I- patients have a single-level fusion.

Type II- patients have multiple, non-contiguous fused segment. Type III- patients have multiple contiguous fused segments.

The anomalies associated with Klippel-Feil Syndrome include:

Scoliosis (60%). Renal abnormalities (35%). Sprengels deformity (30%). Deafness (30%). Mirror motion (synkinesis) (20%). Congenital Heart Disease (15%). Posis, Lateral Rectus palsy Facial nerve palsy. Syndactyly, Hypo plastic thumb. Upper extremity hypoplasia, Neurenteric cyst.

REFERENCES:


eMEDICINE- INTERNET.