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Research Paper

A Rare Angio- Osteo- Hypertrophic Syndrome- Klippel - Trenauny Syndrome

Medical Science

KEYWORDS : KLIPPEL, TRENAUNAY, WEBER, ANGIO-OSTEO, RARE HYPERTROPHY.

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ABSTRACT

Klipel – Trenauny syndrome is a rare syndrome of capillary malformation, venous, lymphatic malformations, associated soft tissue and bone hypertrophy we review a case of 38 yrs old male patient who was referred to our department with multiple port wine nevus, varicose veins, hypertrophy of limbs on one side.

INTRODUCTION:

Here is a rare syndrome which usually presents a triad of port wine nevus, venous and lymphatic abnormalities, associated soft tissue and bone hypertrophy that is klipel - Trenauny syndrome (KTS).

KTS usually involve lower limb, less commonly do occur in upper limb and trunk. There is predilection to gender, race. Presents at birth or early childhood and usually sporadic.

CASE REPORT:

- A 38 year old male patient who was referred to radiology department had multiple visible tortuous vessels , multiple port wine nevus , gigantism of left upper and lower limbs. which were since his child hood .
- Ultrasound, CT, MRI were done.
- Ultrasound reviled multiple tortuous and dilated subcutaneous veins noted involving left upper and lower limbs with increased muscle bulk on ipsilateral side.
- On CT there is increased cortical thickness, width and length of the effected side upper and lower limb bones.
- On MRI increased muscle bulk is noted and multiple dilated subcutaneous veins were clearly visualised.

Fig A &B : CLINICAL IMAGES SHOWING NEVUS INVOLVING LEFT UPPER AND LOWER LIMBS WITH TORTUOUS VESSELS AND LIMB HYPERTROPHY.

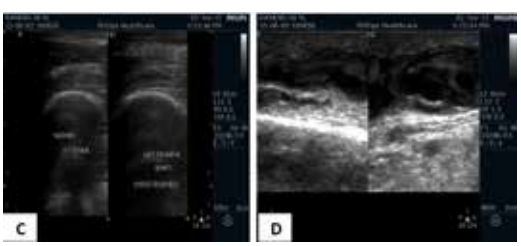

Fig C&D SHOWING ULTRASOUND IMAGES SHOWING INCREASED MUSCLE BULK, FEMUR SHAFT HYPERTROPHY AND DILATED SUBCUTANEOUS VEINS ON LEFT SIDE.

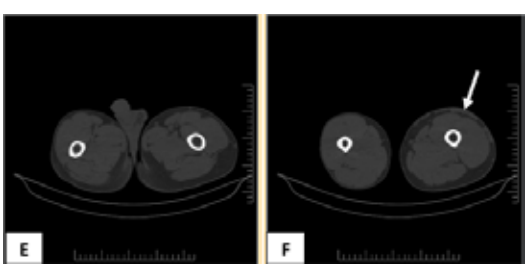

Fig E&F : NECT SHOWING INCRESED MUSCLE BULK BONE THICKNESS ARROW MARK SHOWS DILATED SUB CUTANEOUS VEINS

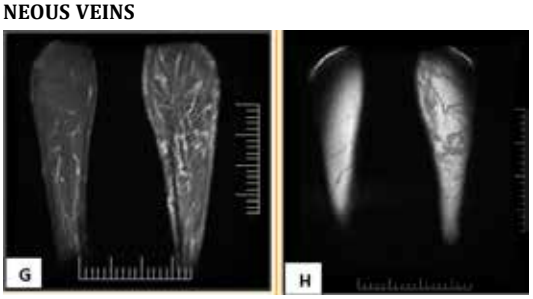

Fig G&H: MRI SHOWING DILATED SUB CUTANEOUS VEINS

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DISCUSSION:

Prevalence of klippel- trenaunay syndrome is approximately 1per 30,000 live births, which is a rare syndrome[1]. Presentation is a classical triad of capillary malformations, vascular anomalies, bone and soft tissue hypertrophy. KTS has a similar, indistinguishable presentation as Parkes-Weber syndrome. However with advanced imaging the differentiation between these two is possible. The differentiation is KTS has low flow arteriovenous malformations (AVMs) and in Parkes-Weber syndrome the AVMs are high flow ones. This differentiation is crucial as high flow AVMs do result in catastrophes such as high output cardiac failure, skin ulceration, and increased limb length discrepancies.[2]

The spectrum of presentation in KTS range from incidental to incapacitating. Ulceration, lymphedema and pain are minor manifestations. However cellulitis, superficial thrombophlebitis which progress to deep venous thrombosis and pulmonary thrombo embolism are grave consequences. Nearly 30% of the cases of KTS are associated with other congenital anomalies like syndactyly and developmental dysplasia of hip.[3,4] one should also be aware of vascular malformations of internal organs which may be source of major bleeds in anemic patients with KTS.[1,5,6]

Etiological hypothesis of the KTS is embryonic mesodermal changes which cause increased angiogenesis leading to increased vascularity, blood flow and tissue hypertrophy.[7] sporadic polygenic mutations is the cause of KTS which is a well-accepted truth.[6] There is another theory of paradominant inheritance which states leathal mutation in a gene, cause survival of heterozygous embryos and failure to survive homozygous embryos. This theory do explain why family links to disease exist and mosaicism that occur in individuals.[5]

Management of KTS is based on specific type and severity of symptoms and never a single pan of treatment recommended. Surgical management is indicated in few cases like varicose veins, conservative management, prior to excision or stripping and these surgical procedures are usually performed after radiological conformation of patent deep venous system. A cut-off difference of more than 2cm was suggested for epiphysiodesis in limb- length discrepancies.[3] Cosmetic correction by laser and sclerotherapy can be made.[4] Debulking operations often fail or worsen symptoms as venous and lymphatic channels gets destroyed. Often treatment is not a definitive in most of the situations, 50% patients re-experience after surgery despite of reported symptom severity improvement in many patients.[8]

DIFFERANTIAL DIAGNOSIS:

Intra uterine ultrasound may diagnose KTWS as early as 15 weeks of gestation, which is by limb hypertrophy and associated subcutaneous cystic lesions. Other features include:

- Foetal hydrops
- Polyhydramnios
- Cardiac failure

Differential diagnosis include:[9]

- Beckwith- wiedemann syndrome
- Proteus syndrome
- Russell – silver syndrome
- Maffucci syndrome
- CHILD syndrome (congenital hemidysplasia with erythroderma and limb defects)
- Neurofibromatosis NF1
- Triploidy

CONCLUSION:

Klippel-Trenaunay syndrome is a rare disease of the vascular and lymphatic system often presenting with port wine nevus at birth. Vascular malformations , hypertrophy of both bony and soft tissues, with hypotrophy being less common. Most cases are difficult to treat due to high rates of recurrence, but individualized intervention can help manage pain and help prevent serious complications.

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