ORIGINAL RESEARCH PAPER

ENT

KEY WORDS: MPNST (Malignant Peripheral Nerve Sheath Tumour), FOP(Fibrodysplasic Ossificans Progressiva,), ROM (Range of Motion), TC ()Tumoral Calcinosis)

UNCOMMON CONDITIONS IN ORTHOPAEDICS: OUR EXPERIENCES

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ABSTRACT

During period ofmay 1998 to October 2018 we came to know about series of uncommon conditions Which never encounterd after that.

1. Tumoral calcinosis is an uncommon condition. Tumoral calcinosis can be classified into 3 types: primary normophosphatemic tumoral calcinosis (the most common), primary hyperphosphatemic tumoral calcinosis, and secondary tumoral calcinosis

Case discription :- A 14 year old tribal boy presented with progressive swelling over right elbow. Radiographs revealed a multilobulated, calcified, progressive mass measuring 11x4x9 cm. Radiographs revealed a multilobulated, calcified, progressive mass measuring 11x4x9 cm. Excision was performed, Histological findings confirmed the diagnosis of tumoral calcinosis Tumoral calcinosis(TC) is an uncommon condition resulting in deposition of calcium in soft tissue especially around large joints. It has been found in patients in Africa but rarely reported from other countries. [1]

Its etiology remains uncertain. Tumoral calcinosis is attributed to an increased calcium phosphate product in the serum, leading to soft-tissue calcification2; the threshold value for precipitation is approximately 5.8 mmol/l.3 Tumoral calcinosis can be classified into 3 types: primary normophosphatemic tumoral calcinosis (the most common), primary hyperphosphatemic tumoral calcinosis, and secondary tumoral calcinosis.4 The first type affects young patients (without any familial history) and is usually a single lesion with low chance of recurrence after excision. The second type is hereditary and usually affects young black men living in the tropics. It is a metabolic disease with decreased fractional phosphate excretion and increased 1,25-dihydroxy-vitamin D synthesis, whereas in proximal renal tubule the response to parathyroid hormone is normal. It affects multiple sites including teeth, vessels, diaphysis and cranium, and recurrence is common. The third type refers to systemic diseases that promote ectopic calcification such as hyperparathyroidism and sarcoidosis

A 14-year-old tribal boy presented with a7-month history of progressive swelling of the right elbow after falling from a swing. The mass was hard and not attached to the arm bones; there was no skin change. The patient had no fever and no pain,numness, or weakness of the arm. The active range of movement(ROM) was full, with flexion from 0ş to 160şand hyperextension of 0ş to 15ş.Radiographs revealed a multilobulated, calcified,progressive mass measuring 11x4x9 cm (Fig. 1).There was no fracture or periosteal change, and the soft-tissue thickness was normal. MRI shows in T2 image inhomogeneous high signal intensity even though there is large amount of calcification. T1 images show inhomogeneous lesion with low signal intensity. The differential diagnoses included tumoral calcinosis and other metabolic calcinosis (such as dystrophic calcinosis, collagen vascular disease, chronic renal disease, hyperparathyroidism). The serum calcium level was 2.43 (normal range, 2.20–2.70) mmol/l, the serum phosphate level was 1.83 (normal range, 0.81–1.94) mmol/l &the serum parathyroidhormone(PTH) level was 2.8 (normal range, 1.6–6.9) µmol/l. As the mass progressively increased in size and malignancy could not be excluded, excision was performed 7 months after presentation. Under general anesthesia, the patient was placed in lateral position with the right shoulder abducted to90ş and the right elbow flexed. A lobulated, yellowish mass with a pseudo-capsule measuring 9x7x4 cm was excised (Fig. 2). It was not attached to surrounding muscles, and some chalky, well-defined material emerged from the surface. Histological findings confirmed the diagnosis of tumoral calcinosis. The mass was transverse by fibrous septa with fibroblastic proliferation.Foreign body giant cells and histiocytic cells were found within the septa. There was no evidence of malignancy.

DISCUSSION

Tumoral calcinosis is a distinct but rare entity in which there is deposition of calcium in peri-articular soft tissue. Our case falls in the idiopathic category since serum calcium and serum phosphorus levels were normal. Hence, it can be grouped under subtype primary normophosphatemic tumoral calcinosis.

In a retrospective study in two institutions over a period of 10 years, amongst 9 patients, the most affected site was the elbow, with the hip coming at second position.5in our case elbow was involved which is common site of affection. Other conditions such as secondary calcinosis, calcinosis universalis, calcinosis circumscripta, soft tissue chondroma, pseudogout, and calcareous tendinitis need to be differentiated from tumoral calcinosis. Plain radiographs in tumoral calcinosis are often diagnostic, showing multiple areas of well-circumscribed, nodular masses with fibrous septa, giving a "cobblestone" or "chicken-wire" appearance. Films exposed with a horizontal beam may show the "sedimentation sign" due to mineral portion pooling dependently, creating a fluid calcium level.6 In our case radiological it was giving cobblestone like appearance posterior to elbow.

Symptomatic treatment is the natural choice, as the cause of the disease is unknown. Only one case of spontaneous regression has been noted.7 Medical treatment with use of calcitonin, bisphosphonates, steroids, phenylbutazone, and radiation therapy have proved to be unsuccessful.8Complete surgical excision of tumoral calcinosis is considered to be the optimum treatment. The recommended management for tumoral calcinosis is surgical excision. Complete excision of mass is required to prevent recurrence. Medical treatment using agents that decrease serum phosphate levels have limited use in the management of tumoral

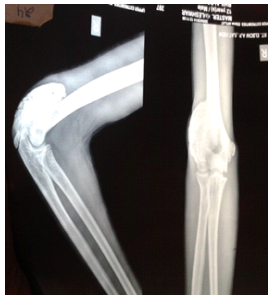

Fig.1 xray elbow tum.calcinosis

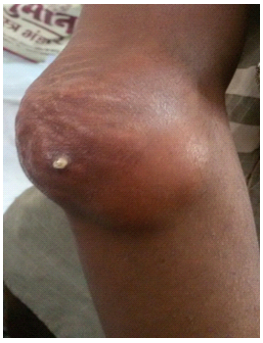

Figure 2:clini.pic.tum.calcinosis

2.MALIGNANT PERIPHERAL NERVE SHEATH TUMOUR

ABSTRACT

A 18 year male resident of distt. Rajnandgaon, came at Dr. B.R. Ambedkar hospital with complaints of painless swelling over left hand, of size 9cm./7cm. over webspace of little and ring finger .

He was investigated with X-ray, MRI & FNAC . then treated with wide excision of tumour with amputation of little finger from MCP joint and skin grafting , supplemented with adjuvant radiochemotherapy .

Histopathological examination reveal Malignant Peripheral Nerve Sheath Tumour (MPNST).

INTRODUCTION

Malignant peripheral nerve sheath tumour(MPNST) is a rare variety of soft tissue sarcoma of ectomesenchyma .Tumour arises from major/ minor peripheral nerve branch or sheath of peripheral nerve, tumour many a times associated with neuro fibromatosis .Gross ,histopathological and immunohistochemical examination used for diagnosing these tumours. It is multifocal in nature .It is gross fusiform tumour in relation to microscopic features of spindle cell with fascicular pattern and varying degree of mitosis and necrosis sometimes tumour in subendothelial zone , neoplastic cell herniates into vessels lumen and proliferates in its wall MPNST also known as malignant schwannoma derived from Schwann cell or pluripotent
Cells of neural sheath,epitheloid or heterogenous component can be observed in15% of MPNST.,latter include rhabdomyoblast,osseous,cartilaginous differentiation and rarely Smooth muscle, glandular and liposarcomatous component have been reported.

Surgery is the mainstay of treatment.Operativesteps:Brachial block was given prior to surgery.preperation was done for excision of finger along with tumour as the tumour was badly adhered with finger and skin grafting done with graft taken from thigh which were kept ready ,with pt.kept palmer aspect upward, bleeding was checked with cautry with explained prognosis regarding sacrifice of affected finger.pt. stood surgery very well. No perioperative complication occur.

supplemented with adjuvant radiochemotherapy ,though this tumour is biologically aggressive in nature.

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DISCUSSION

MPNST is a rare variety of tumour with incidence of 1/100000 population A combination of gross finding with immunohistochemical study is commonly used to diagnose a case of MPNST .However it is not always possible to demonstrate the origin of cell especially when it arise from sheath. Tumour displays fascicles of spindle cell Woven into herring bone pattern with varying degree of mitosis The tumour is more common in males than females The survival rate varies upto 8 years.
MPNST has highest rate of any sarcoma,

constitutes significant proportion of soft tissue sarcoma.

Incidence of MPNST in tumour prone genetic condition

Disease ;	risk ;
1. neurofibromatosis	1252
2.schwannomatosis	181
3.fam.adenom polyposis	477
4 triton tumour	221

MPNST;	SEX ratio,M:F	Mean age
52	22:30	31(13-77)
3	2:1	26(17-45)
0	0	- -
3	2:1	32(1-33)

A combination of clinical ,pathoimmunohisto-chemistry helps in diagnosing the tumour.Post operative radiotherapy has definite role in both disease and overall survival.Tumoralcalcinosis is a rare entity. Hence, the surgeon must be aware of this possibility. It needs thorough evaluation to exclude other causes of soft tissuecalcification. Once the diagnosis is established, completesurgical excision is the only option for cure and preventionof recurrence.

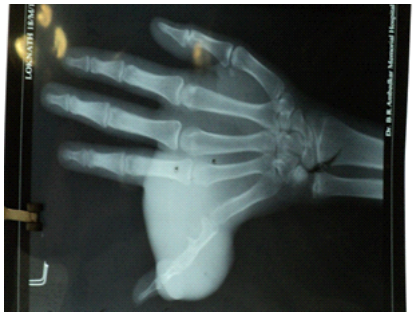

Fig.3xray hand MPNST

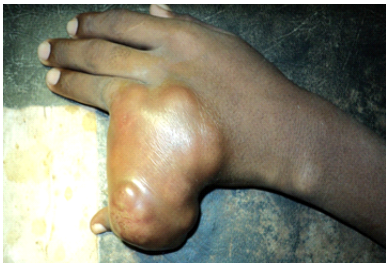

Fig.4 clin.pic MPNST

3Eccrine Acrospiroma Hand,An uncommon condition

Abstract- Presenting a case of ECCRINE ACROSPIROMA HAND; An uncommon condition in a 33 years old man complaining of painless swelling circular 4.8 cm x 2.6cms over leftmiddle finger since 4 months.This benign tumour is distinctive sweat gland tumour occurs as asingle mass in the skin and solid/cystic,the colour varies from that of surrounding skin to red or reddish blue and covering skin may besmooth or thickened and verrucous .This tumour are rarely painful andserous or hemorrhagic fluid may drain spontaneously from them. Theymay recur but rarely undergo malignant change.Histologically, the acrospiroma is readily differentiated from other sweatglandtumour but frequently confused with lesions of metastatic renal cellcarcinoma and with lesions of a squamous cell carcinoma .On the basisof histological studies we believe that cells of tumour mimic those of eccrine sweat gland and have designated the entity asECCRINEACROSPIROMA.

Introduction- The acrospiroma appears as a single ,solid or cystic mass that Protrudes from dermis .The diameter of tumour ranges from 0.5 to 10.5cm x1cms .The median age of primary tumour varies 3-93 years, median age being 33 yrs.,predominantlyMales but can occur at any part of the body and usually painless in nature not associated with trauma.

Grossly tumour appears to be delineated from the surrounding tissue but were not encapsulated ,usually firm and solid but some

contain cystic foci filled with serous to gelatinous ,clear to hemorrhagic material.Figure 5: xray eccrine acrospiroma Histologically , tumor are circumscribed ,noncapsulated, multilobular mass lying in the upper or the middle dermis.The lobules were circular or irregular and had scalloped borders .The epidermis covering of most of tumour was acanthotic.The tumour composed of biphasic epithelial cells surrounded by variable amount of stroma in all phases cells were round ,fusiform or polyhedral,nucleus was oval to round with fine reticular chromatin and a distictnucleolus.the lumen contained an amorphous eosinophilic homogenous material ,sometimes contains haemorrhagicfoci.The eccrine ducts near the tumour showed proliferation of cells within the tumour, The melanin pigment is present in macrophages of the tumours..The tumour is designated as an eccrine acrospiroma because spiroma means adenoma of sweat gland and acro indicates the topmost or end.The eccrine acrospiroma is a benign tumour arising from myoepithelial cells of the sweat gland . may reoccur locally after excision.Usually located on planter aspect of foot and rarely over hand .Excision of tumour was done under ring block of middle finger, haemostasis achieved by cautry.and complete excision done.patient sustained operation well . excised matter was send for biopsy and patient was discharged in the evening with oral antibiotics and analgesics with good results. [Figure 5&6]

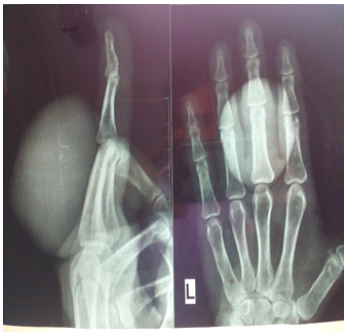

Figure 5: xray eccrine acrospiroma

Fig6 clini.pic eccrine acrospiroma

4.Stoneman's disease,an uncommon disease

Fibrodysplasia ossificans Progressiva is a rare and disabling genetic condition characterized by congenital skeletal malformations and progressive heterotopic ossification in humans with no ethnic, racial, gender, or geographic predilection. Diagnosis of this condition can be made clinically in presence of radiographic evidence of heterotopic ossification along with symmetrical malformations of the great toes. The course of disease is unpredictable often progresses from early childhood, become immobile and confined to a wheelchair by their twenties. Survival beyond the third decade is uncommon. We hereby report a case of Fibrodysplasia ossificans Progressiva in a 7˝ old girl. Fibrodysplasia ossificans Progressiva (FOP) is also known as Myositis ossificans progressiva, (Stoneman disease, Munchmeyer's disease) is an exceptionally rare autosomal dominant disorder of connective tissue characterized by congenital malformations of the great toes and progressive heterotopic ossification in characteristic extraskeletalsites. The likely incidence is about 1 in 2 million with no ethnic, racial, gender, or geographic predilection. The diagnosis is based on the clinical findings and radiological demonstration of the skeletal malformations. We hereby report a case of FOP in a 7˝ old girl from Chhattisgarh.

Case History:

A 7˝year old female child admitted in our hospital with complaint of inability to raise both shoulders. She had painless swelling over lower back on both the sides 6 month back which gradually regressed in size and disappeared. There was no history of trauma, systemic illness or prior hospitalization. Perinatal history and family history were insignificant. She was well and active. Her growth and developmental milestone was appropriate for the age.

On examination, there was the drooping of the left shoulder. There was a single bony hard subcutaneous swelling overlying right scapula. The swelling was non-tender and no signs of inflammation were present. There was a significant restriction of abduction and internal rotation of both the shoulders. Partial restriction of all movements of the spine and restriction of flexion of the neck was also present. Physical examination showed stiffness in the entire spine, affecting from cervical to lumbar region. Affected muscle was stony hard in consistency, immobile and painless to palpation, and showed no inflammatory signs. Clinodactyly of both little finger and slightly shortening of both great toes were present. Her Hearing assessment reveals a normal study.

Radiographs revealed hypoplasia of proximal phalanges of great toes bilaterally, ectopic ossifications in bilateral axillary region, both sides of the neck and in the Para vertebral muscles. There is calcification of entire right sternocleidomostoid muscle (figure 1 & 2). Laboratory tests such as blood cell count, serum calcium, alkaline phosphatase, parathormone, vitamin D3, C-reactive protein, rheumatoid factor, and complete urine examination showed normal values. Considering these results, and analyzing the signs and symptoms presented by the patient, the clinical diagnosis of Fibrodysplasia Ossificans Progressiva was made.

DISCUSSION:

Myositis ossificans (MO) is an extra-osseous non-neoplastic growth of new bone. The two main recognized MO subtypes are known as traumatic myositis ossificans which occurs following trauma (uncommon in children) and fibrodysplasia progressive ossificans (FOP). FOP is a rare, fatal, inherited disorder causing fibrosis and ossification of muscles, tendons and ligaments1,2.Pathogenesis of traumatic myositis ossificans is still remains unclear. Most of the researchers believe that repetitive small traumas (not recognized by the patient), infection, inflammation and ischemia may be the underlying factors that contribute to non-traumatic MO 3.

FOP occurs in approximately 1 in 2 million people.FOP is a genetic disease inherited autosomal dominant pattern in most of the cases while remaining cases are caused by a spontaneous new mutation in the ACVR1 gene. This mutation causes a deregulation of the bone morphogenetic protein signaling pathway4,5Previous studies have demonstrated that there was no ethnic, racial, gender or geographic predisposition for the development of the disease 6,7.

Recognition of the most characteristic deformity microdactyly of both halluces due to a single phalanx in association with rapidly changing swellings begins during the first decade of life, at 4 years on average, (in the present case it was 7˝ years) and progress until being ossified helps to make a diagnosis. Swellings typically affect neck and upper back making them stiff, as in our case. The diagnosis of FOP is therefore based upon history, clinical and radiological findings.

The bilateral great toe anomaly present from birth, reported in 79 to 100% of patients. Deafness has been reported in up to one fourth of the cases. Malignancy is the most common misdiagnosis with up to 1 in 3 cases being mistaken as a tumor. The soft tissue trauma can induce rapid ossification of the affected area; therefore, biopsy of calcified nodules is to be avoided if the

diagnosis of FOP is clear on clinical and radiological grounds.

Soft tissue ossifications are the characteristic radiographic features of FOP (figure 3). Bone scintigraphy with 99mTc-MDP may demonstrate early the heterotopic ossification and aid in the assessment of the extent and progression of the disease8. Laboratory analysis, biochemical values are usually found to be normal as in our case, although we could not carry out bone scans and mutation study. Our case demonstrates the classical presentation and features of FOP. No effective medical therapy is known for fibrodysplasia ossificans progressiva; bisphosphonates and corticosteroids are only drugs have some benefit during acute phase9. Gene therapy may have a certain role.

The course of disease is unpredictable often progresses from early childhood, become immobile and confined to a wheelchair by their twenties. Survival beyond the third decade is uncommon, as severe restriction of the chest wall results in cardiorespiratory failure.

Myositis ossificans progressiva is very uncommon hereditary disorder and must be consider in those cases where extraosseous nonneoplastic new bone formation is seen. Clinical suspicion of fibrodysplasia ossificans progressiva early in life on the basis of malformed great toes can lead to early clinical diagnosis, confirmatory diagnostic genetic testing, and the avoidance of harmful diagnostic procedures. [Figure 6 & 7]

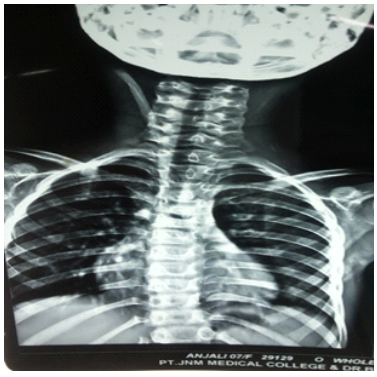

Figure 6 XRAY; FOP

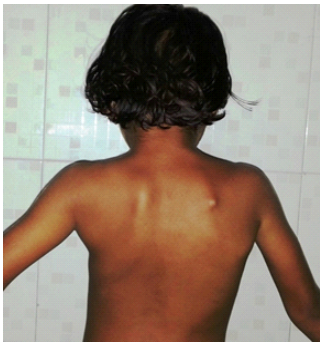

Figure 7clin.pic FOP

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