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RESEARCH ARTICLE

CASE REPORT : A RARE CASE OF CUTANEOUS DESMOID-TYPE FIBROMATOSIS

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Abstract

Desmoid fibromatosis (DF) is a rare, locally aggressive connective tissue malignancy primarily affecting musculoaponeurotic tissues, representing less than 3% of all neoplasms. This case report presents a 30-year-old male with cutaneous desmoid fibromatosis, a rare variant characterized by a painless, firm mass in the left shoulder region. Histopathological, immunohistochemical, and molecular analyses confirmed the diagnosis of desmoid cutaneous fibromatosis, with positive findings for nuclear beta-catenin. Management involved a multidisciplinary approach, with a conservative strategy due to the tumor's size and location, emphasizing regular monitoring for recurrence. Differential diagnosis considerations included dermatomyofibroma and scar tissue. Treatment options for DF vary, with a shift towards observation as the primary strategy, particularly in cases where surgery may result in significant functional impairment. Medical treatment or surgery may be considered in symptomatic or progressive cases. Close surveillance and follow-up imaging are crucial for early detection of disease progression. cutaneous desmoid fibromatosis presents diagnostic challenges, highlighting the importance of early identification and biopsy to prevent misdiagnosis.

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Introduction:-

Desmoid fibromatosis (DF) is a locally aggressive, deep-seatedconnective tissue malignancy developing in musculoaponevrotic tissues. also known as aggressive fibromatosis, deepfibromatosis [1][2].

It represents less than 3% of all neoplasms and 0.03% of all soft-tissue tumors[1]. The yearly incidence is roughly 2-4 cases/million, peaking in the ages of 6–15 and 25–35[3].

Though it can affect almost any portion of the body, it usually affects the extremities,abdominal wall, and abdominal mesentery[4]. DF has a strong tendency to recur even though it has no potential to spread. The clinical appearance of these uncommon tumors varies greatly, and their natural history is uncertain.Surgical resection was the usual management strategy for DF in the past. But new developments in DF's molecular cytogenetics and tumor biology have shifted the management of this condition.

This case reports a patient withcutaneous desmoid fibromatosis which is a rare variant . The report emphasizes the challenges associated with the management of this rare tumor and highlights the importance of a multidisciplinary approach for optimal patient outcomes.

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Presentation Of The Case

We present the case of a 30-year-old male patient who presented with a painless, firm mass in his left shoulder region. The mass had been gradually increasing in size over the past six months. Physical examination revealed a palpable, non-mobile mass measuring approximately 5 cm in diameter, with retractile hyperpigmented surface .Ultrasonography showed soft-tissue mass involving the dermis and the subcutaneous tissue ,biopsydemonstrate A lesion located in the deep dermis and especially the hypodermis, made up of spindle-shaped cells which seem to respond to fibroblasts and myofibroblasts arranged in long divergent bundles, on the whole not very cellular and often containing undulating collagen.The cells do not show any clear cytonuclear abnormalities and mitosis are not visible.The nuclei are poorly stained and have one to three small nucleoli.The lesion contains small vessels sometimes surrounded by clear halo , It is punctuated by a few lymphocytes.in complementary immunohistochemical study , the lesion cells showed diffuse positivity for smooth muscle actin and beta-catenin, and were negative for CD34 and PS100.In order to better characterise this lesion, a mutational panel targeted by next-generation sequencing was carried out.sequencing revealed the presence of the C. 121A> G mutation in exon 3 of the CTNNB1 gene. These histopathological, immunohistochemical and molecular results,suggested a diagnosis of desmoid cutaneous fibromatosis

A multidisciplinary team comprising desmatologists ,surgeons and oncologists, was involved in the management plan. Due to the size and location of the tumor, a conservative approach was adopted, with regular clinical and radiological monitoring. The patient was enrolled in a surveillance program, including regular imaging studies to monitor tumor growth and potential signs of recurrence ,MRI after two years follow-up ,did not show evidence of volume increasing

Discussion:-

In the International Classification of Diseases (ICD) DF is classified as D48.According to the World Health Organisation (WHO),DT is a "clonal fibroblastic proliferation that arises in the deep soft tissues and is characterised by infiltrativegrowth and a tendency toward local recurrence but aninability to metastasise"[5] .although the exact etiopathogenesis of DF is unknown, it is thought to be complex[6][7]. Desmoid fibromatosis can occur in a familial or sporadic cases . The etiopathogenesis has been linked to oral contraceptive use, pregnancy, and trauma.[6][7] Desmoid fibromatosis is most likely to arise at surgical scar sites, particularly those following cesarean sections and intraabdominal resections.

There are two forms of fibromatoses: superficial and deep [8]. Knuckle pads, infantile digital fibromatoses, plantar fibromatoses (Ledderhose's disease), penile fibromatoses (Peyronie's disease), palmar fibromatosis (Dupuytren's contractures), and infantile digital fibromatoses are examples of superficial fibromatoses[7].deep fibromatoses may affect any site but is commonly seen in the extremities, abdominal wall, and abdominal mesentery [9], it is a locally aggressive, deep-seated connective tissue malignancy developing in musculoaponeurotic tissues, But in our case it is centred in the dermis and hypodermis, suggesting that desmoids can sometimes develop in the skin[10].

While expression of nuclear beta-catenin can be seen in superficial fibromatosis, mutations in CTNNB1 or APC, which are common in desmoids, have not been identified in superficial fibromatosis[11][12]. Moreover, cases of superficial fibromatosis are essentially confined to four anatomical sites: palms, soles, knuckles and penis.which allow us to exludethe superficiel fibromatosis diagnosis , but several differential diagnosis should be considered , especiallydermatomyofibroma, scar, and leiomyoma. A dermatomyofibroma is a diagnostic consideration, particularly due to subcutaneous involvement and preservation of epithelial/adnexal units, as seen focally in this case. In dermatomyofibroma, the spindle cells typically grow in fascicles parallel to the epidermis. Furthermore, the cells of dermatomyofiboma have not been reported to express nuclear beta-catenin, although only a small number of published cases have reported this marker[13][14][15].While the lesion shows some histologic resemblance to a hyperplastic scar, the lack of a history of preceding trauma and absence of vertically-oriented capillary-type vessels argues against this possibility. Interestingly, nuclear positivity of beta catenin has been reported in the context of scars[16][17].The typical sign of desmoid-type fibromatosis is a slow expanding, which may or may not be painful[13]. and there biological behavior is often unpredictable, certain lesions may exhibit more aggressive activity. Local spontaneous recurrence is common, although metastasis or sarcomatous transformation have not been observed in cases of desmoid-type fibromatosis[18].Immunohistochemical stains are helpful and should be perform in every case. Immunohistochemical staining usually showpositive for nuclear B-catenin, vimentin, cyclooxygenase 2, tyrosine kinase PDGFRb, androgen receptor and estrogen receptor β but negative for desmin, S-100, h-caldesmon, CD34, and c-KIT[19][20].

The management of DF is challenging, and treatment options depend on the location, size, and aggressiveness of the tumor, as well as the patient's age and overall health. Treatment options include surgery, radiation therapy, systemic therapy, and observation , which became throw the last decade the most adopted strategy [21]. with most centers moving away from primary radical surgery towards a front-line "watch-and-wait" policy[22][23].

Medical treatment should be considered next if the disease is symptomatic or progressive during surveillance. Surgery should only be employed when medical treatment fails to achieve radiological stabilization or clinical improvement, and if surgery is possible without significant functional impairment[23][24].If surgery will lead to substantial disability, chemotherapy or radiotherapy may be a preferred option[22].A decision towards an active treatment should be postponed until the occurrence of subsequent progression or increase of symptoms burden, assessed with at least two further assessments and possibly not before one year from diagnosis the aim of systemic treatment is to slow or stop growth of fibromatosis as well as improve disease-related symptoms such as pain[25].Follow-up imaging should be performed at 4-8 weeks to avoid missing rapidly progressing disease, especially for lesions that are not easily palpable or that are located at critical sites. If MRI is stable, follow-up is 3-monthly for the first year, 6-monthly up to the 5th year, and yearly thereafter. For patients on medical treatment, imaging is generally performed at 3-6 month intervals, depending on symptoms and site of disease[26].

In summary, cutaneous desmoid fibromatosis is an extreamly rare type of fibrous tumors which can create a huge confugion for clinicians. Therefore, it is suggested that clinicians should carefully identify such cutaneous tumor and earlier conduct biopsy to avoid wrong diagnosis.

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