ORIGINAL RESEARCH PAPER
Volume - 10 | Issue - 02 | February - 2021 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

ORBITAL LIPOFIBROMATOSIS

Ophthalmology

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ABSTRACT

The purpose of the study was to report a case of orbital lipofibromatosis in an adult patient. A 44 years old male patient presented with history of gradually expanding mass in superior quadrant of left orbit since 1 year. Contrast Computed Tomography (CECT) of orbits showed left superior rectus muscle thickening with associated periorbital fat stranding and preseptal edema. Positron Emission Tomography (PET Scan) was done to rule out any lymphomatous involvement. Histopathological examination revealed that the lesion consisted of mature adipocytes with fibrous tissue, being consistent with a diagnosis lipofibromatosis.

KEYWORDS

Lipofibromatosis; Orbital tumour; Soft tissue tumour

INTRODUCTION

Lipofibromatosis is a rare infiltrating, benign tumour of fatty tissues mostly seen in children which was first described by Fetsch et al[1]. It is usually presented with a soft tissue mass involving upper and lower extremities. Involvement at other sites (i.e., abdominal wall, chest wall, mandible, neck and orbit) has also been reported[2,3,4]. Herein, we describe the rare case of lipofibromatosis involving the orbit of an adult.

CASE REPORT

A 44-year-old diabetic male patient presented in outpatient department of ophthalmology with chief complaint of slowly growing and painless proptosis since 1 year. There was a history of blurring of vision in the left eye. Past medical history revealed that patient was diagnosed as orbital psuedotumour of left orbit for which oral corticosteroids were given for 1 month. On examination, the visual acuity in his left eye was 6/36. Movements of the left eye were limited in all gazes but more pronounced in depression. There was non-axial proptosis of the left eye, which was displaced upwards and outwards. On palpation, a firm, non-tender mass was present in the superior part of left orbit which was resistant to retropulsion. Slit lamp examination showed mildly congested conjunctiva in left eye. Cornea was clear in both eyes. Pupil was round, regular and reacting to light in right eye but relative afferent papillary defect was present in left eye. Fundoscopic examination of both eyes showed normal optic disc, blood vessels and macula.

Contrast computed tomography of the orbit showed heterogenous mass in left orbit with thickened left superior rectus muscle. Positron Emission Tomography (PET Scan) was done to rule out any lymphomatous involvement and showed metabolically active thickened left superior rectus and medial rectus muscles. The radiological findings suggested a diagnosis of left orbital pseudotumour and patient was started with intravenous methyl prednisolone (1gram/day) for 5 days. Lesion was partially responsive to treatment. Patient visual acuity improved to 6/9 and pupil became round, regular and reactive to light in left eye.

Histopathological analysis showed fragments of fibrocollagenous tissue containing lobules of mature adipocytes, blood vessels and skeletal muscle fibres. Bundles and fascicles of fibrous tissue transversing in the septa between adipose tissue and around skeletal muscle were seen. The cells were spindle shaped with plump and stellate nuclei. There was no myxoid change. No significant mitosis, pleomorphism or any evidence of malignancy was noted.

Immunohistochemistry showed spindle cells were strongly positive stained for CD34 and weak positive stained for Desmin, SMA and β-catenin.

Based on the histolopathology and immunohistochemistry, a diagnosis of lipofibromatosis was made and patient was planned for chemotherapy (Injection Rituximab, 6cycles) followed by radiotherapy.

DISCUSSION

Lipofibromatosis is a rare, benign paediatric soft tissue tumor previously designated as infantile fibromatosis of non-desmoid type. The etiology remains unknown but different hypothesis have been proposed, including embryonal or foetal cell malformation, and paracrine growth regulation disorder.[5,6] Cytogenetic abnormality with a three-way t (4:9:6) translocation has also been reported.[7]

Lipofibromatosis has been described from birth to the early second decade. The tumours are slow-growing and present with no pain. There is a male predominance with a male to female ratio of 2:1[8]. It is moslyt seen in hands and feet and is slightly less common in the thigh, trunk, jaw, orbit, head and neck[1-4]

.The diagnosis of lipofibromatosis is mainly based on standard histopathological analysis. On histological examination, the tumour is characterised by alternating lobules of mature adipocytes and fascicles of variable cellular but bland fibrous tissue. The fibroblasts are spindled in shape with ovoid nuclei, no cytologic atypia and minimal mitotic activity. Unlike fibromatosis, lipofibromatosis generally does not demonstrate solid, sheet-like fibrous growth. Commonly, lipofibromatosis exhibits staining of the spindle cells with CD34, CD99, smooth muscle actin. BCL-2, S-100 protein, muscle specific actin and EMA may be occasionally expressed.

Surgical resection is the elective treatment for lipofibromatosis[9]. If the lesion is incompletely excised, local recurrence may occur. In this case, surgical excision of the lesion was not possible, so patient was planned for chemotherapy followed by radiotherapy if required and patient has been kept under follow up.

Figure 1 & 2: Firm mass in left orbit; Clinical presentation and Axial CT Scan

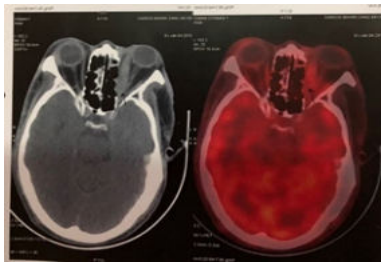

Figure 2: Axial CT scan showing mass in left orbit with thickening of left superior rectus muscle

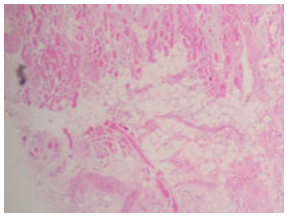

Figure 4: Microscopic view showing ,fibrous septae separating adipose tissue on H&E stain.

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