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A Rare Combination of Neurofibromatosis and Fibrous Dysplasia.

Roshni J*, I Venkatraman, and Ravindran.

Department of Radiodiagnosis, Sree Balaji Medical College & Hospital, Chennai, Tamil Nadu, India.

ABSTRACT

Neurofibromatosis and fibrous dysplasia were both described by von Recklinghausen. There are speculations if neurofibromatosis and fibrous dysplasia are different manifestations of the same disease or if these conditions are related in someway. Here we report a patient with facial swelling (which turned out to be Neurofibroma), expansion with ground glass density of maxillary sinus walls and café au lait spots on the back. We report this case as neurofibromatosis coexisting with fibrous dysplasia is very rare and only few cases have been reported worldwide.

Keywords: Neurofibromatosis, Fibrous dysplasia, Craniofacial, Computed tomography (CT)

Corresponding author

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INTRODUCTION

Neurofibromatosis (NF) is an autosomal dominant disorder. NF1 occurs in approximately 2,500 -3,300 live births [1]. The term neurofibromatosis is used for a group of genetic disorders that affect the cell growth of neural tissues. It is a neurocutaneous condition that can involve any organ. Fibrous dysplasia (FD) is a congenital, non-hereditary, progressive, skeletal disorder in which normal bone is replaced by fibrous and osseous tissue [2].

Case Report

A 20 years old male patient came with complaint of swelling in the face involving bilateral cheeks that persisted for 10 years. The swelling has gradually increased in size and is now obscuring the field of vision. On palpation the swelling was of normal temperature, non-tender, firm in consistency and skin over the swelling was pinch able. On general examination, patient had café au lait spots over the trunk.

Plain and Contrast enhanced computed tomography (CECT) of face revealed a large soft tissue density lesion in the right infratemporal fossa causing widening and scalloping of the right pterygopalatine fossa with extension into the right orbit and pre maxillary region causing right proptosis [Figure 1]. Soft tissue density lesion was seen in the inferior aspect of left orbit with extension into the widened left infraorbital canal and foramen. Anteriorly the lesion was seen extending into the left premaxillary region. Both the lesions show homogenous moderate contrast enhancement on the delayed contrast images [Figure 2 & 3]. Neurofibroma was considered as possibility. Expansion and ground glass density was seen involving floor and walls of maxillary sinuses, bilateral ethmoid sinuses and bilateral inferior and middle turbinates with soft tissue density filling the narrowed cavities of bilateral maxillary sinuses and nasal cavity with extension of soft tissue density into the nasal cavity [Figure 1]. Possibility of fibrous dysplasia was considered. Histopathology of the soft tissue swelling in the cheek was consistent with neurofibroma.

Figure 1: Axial CT – Bone window

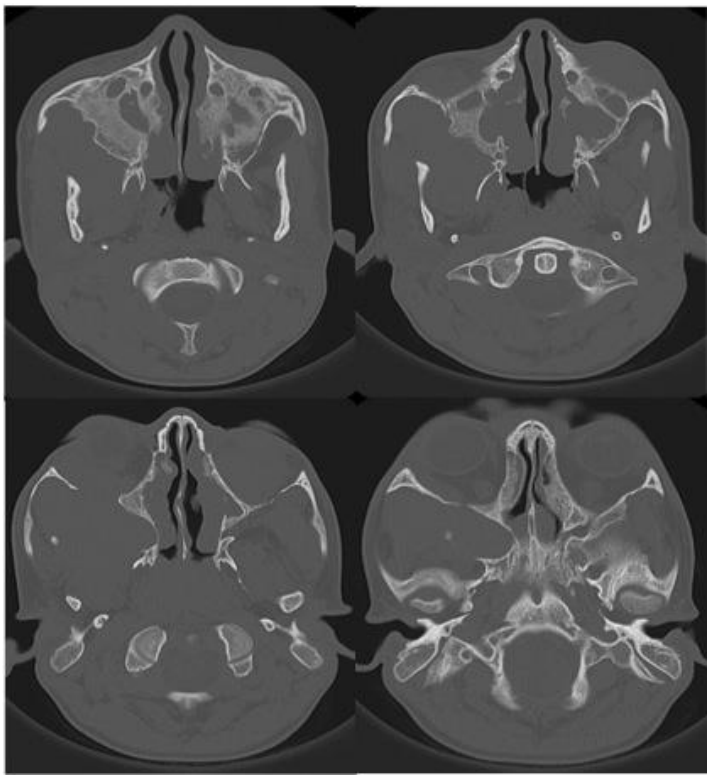

Figure 1: Widening and scalloping of the right pterygopalatine fossa with extension into the right orbit and pre maxillary region. Expansion and ground glass density was seen involving floor and walls of maxillary sinuses, bilateral ethmoid sinuses and bilateral inferior and middle turbinates.

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Figure 2: Plain and CECT axial – Soft tissue window

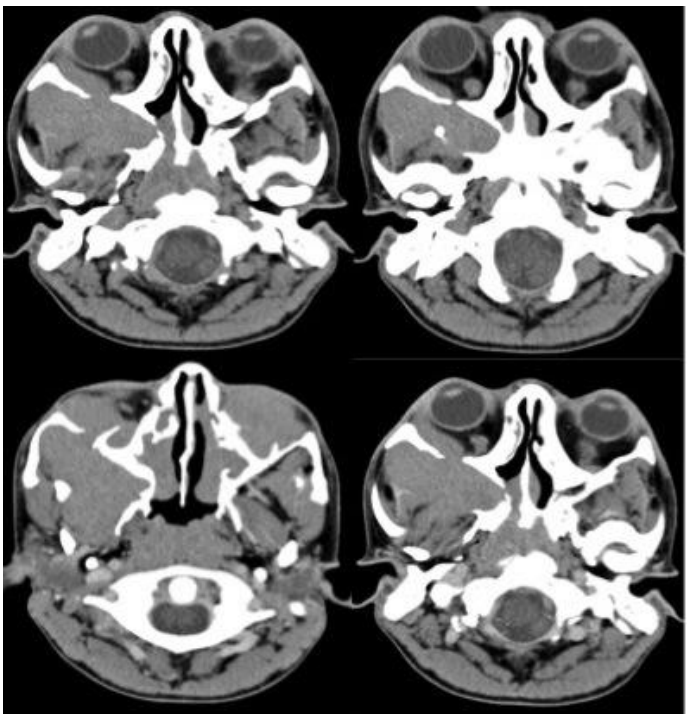

Figure 3: CECT coronal – Soft tissue window

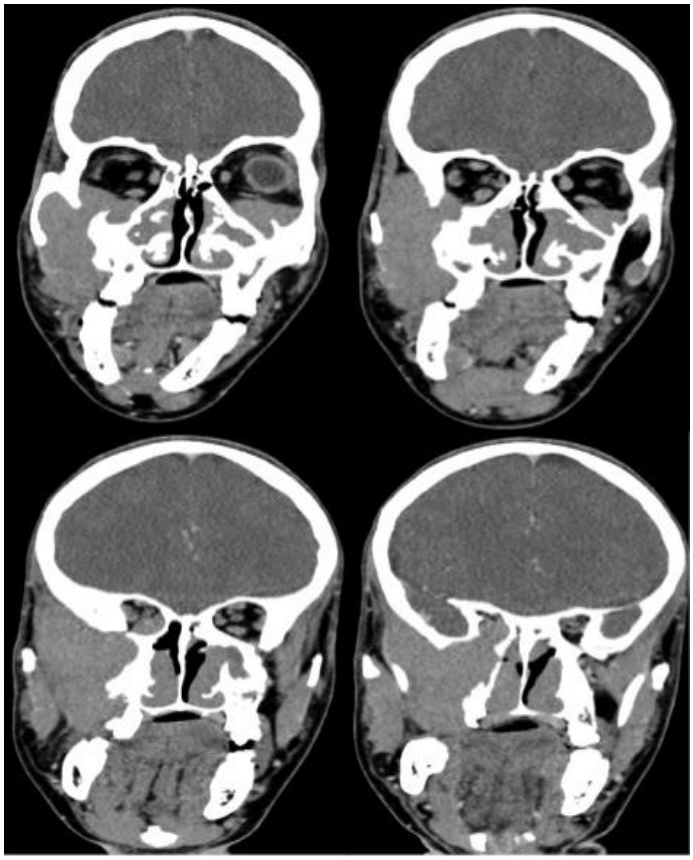

Figure 2 & 3: Soft tissue density lesion was seen in the inferior aspect of left orbit with extension into the widened left infraorbital canal and foramen. Anteriorly the lesion was seen extending into the left

premaxillary region. Both the lesions show homogenous moderate contrast enhancement on the delayed contrast images.

DISCUSSION

Neurofibromatosis and fibrous dysplasia were described by von Recklinghausen [3].

The onset of FD is most often seen in the first decade of life, and usually ceases its progressive course at skeletal maturation, whereas NF does not manifest before the second decade [4]. In the present case, both NF1 and polyostotic FD manifested almost at the same time, in the second decade of the patient's life. During embryogenesis, a regulator gene mutation with variable penetrance is responsible for both these diseases [5].

Neurofibromatosis is inherited as an autosomal dominant disorder, but there is a high rate of new mutations.

Two major subtypes of NF are Neurofibromatosis type 1(NF1), referred as peripheral neurofibromatosis and Neurofibromatosis type 2, referred as central neurofibromatosis, with a third variant known as segmental neurofibromatosis - limited to a single body region.

Pigmented lesions are a common manifestation in NF1. These lesions usually appear during the 1st year of life or are present at birth, either as cafe au lait spots or as freckles [6 -10]. Cafe au lait spots are hyper pigmented macule that vary in color form light brown to dark brown. They have smooth or irregular borders. They appear anywhere on the skin, but they are less common on the face [6].

Bone involvement in NF1 may be due to external resorption and internal osteolytic defects. In NF1 bone malformations like kyphoscoliosis or pseudoarthrosis may appear, and temperomandibular joint may be involved [8,10,11]. Skeletal involvement is seen in almost 40% of patients with NF1, of which scoliosis is the most common skeletal pathology [9,11,12].

Involvement of oral cavity is seen in 66-72% of the cases of NF1. The most frequent finding of lengthening of fungiform papilla is seen in 50% of cases. In 2% of oral neurofibromatosis patients, oral neurofibromas can be seen. Neurofibromas can appear in every tissue, soft or hard, in the oral cavity [6,9,10,13,14]. NF1 patient may also show facial disfigurement due to hypo or hyperplasia of maxilla, mandible, malar bone, and temporomandibular joint.

Histologically, neurofibromas are composed of a mixture of Schwann cells, perineural cells and endoneural fibrobalsts, and they are not capsulated [8,14,15]. Schwann cells account for about 36-80% of lesional cells. They form the predominant cellular type and usually have widened nuclei with an undulated shape and sharp corners [15].

Fibrous dysplasia is a congenital, non-hereditary, progressive, skeletal disorder in which normal bone is replaced by fibrous and osseous tissue. [2]. It may affect one (monostotic) or multiple bones (polyostotic). Patients have functional and cosmetic complaints like facial swelling or asymmetry, facial pain and paresthesias as well as compressive symptoms like nasal obstruction, sinusitis, hearing loss and visual disturbances [16]. The mixture of fibrous and osseous elements in FD causes the characteristic homogenous “ground glass” appearance with ill-defined borders by radiographic study. The affected bone is sometimes widened, and it may appear sclerotic, lytic or mixed. There is no periosteal reaction or soft tissue involvement. CT scan is a better radiographic tool for assessing the extent, especially in cases of suspected optic canal involvement. Radionuclide bone scintigraphy is useful to assess the extent of the disease. Diagnosis of FD is based generally on the clinical ground and typical radiographic findings. Bone biopsy is typically reserved for cases suspicious of malignant change.

Craniofacial fibrous dysplasia is different and it ignores suture lines; more than one bone is usually involved. The radiologic appearance of fibrous dysplasia appears as a lucent area with a sclerotic rim. In the skull base and facial bones, fibrous dysplasia manifests as marked sclerosis and bone thickening.

Treatment with bisphosphonates is found to be beneficial in FD, resulting in rapid pain relief and normalization of bone turnover [17]. The management of NF1 is currently focused on genetic counseling and esthetic treatment of specific lesions, usually through surgery [18].

CONCLUSION

A possible relationship between NF and FD was found through a literature search, and has been substantiated in our case. An understanding of this relationship would be more than just academic importance, and might be of therapeutic value. If this is so, all NF patients should be routinely reviewed for the above associated diseases, which could be rewarded with an early diagnosis and better prognosis.

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