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Abstract

Neurofibromatosis and fibrous dysplasia show the presence of cafe-au-lait spots, bone lesions, and endocrinopathies.

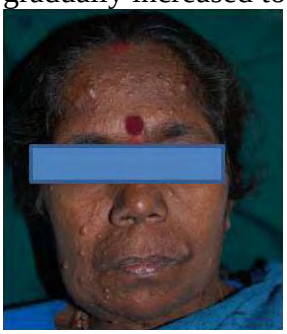
There has been speculation whether neurofibromatosis and fibrous dysplasia are different manifestations of the same disease or if these conditions are in some way related.

We provide a case of whether neurofibromatosis and fibrous dysplasia complicated by hyperparathyroidism and osteoporosis.

Introduction

Case report

- A 45-year-old female patient complaint of **swelling** on the right body of the mandible that had persisted for **6 months**. The swelling was initially small, and gradually increased to the present size of **3 X 4 cm**.



On palpation, the swelling was of normal temperature on touch, tender and firm inconsistency, and the skin over the swelling was pinchable.

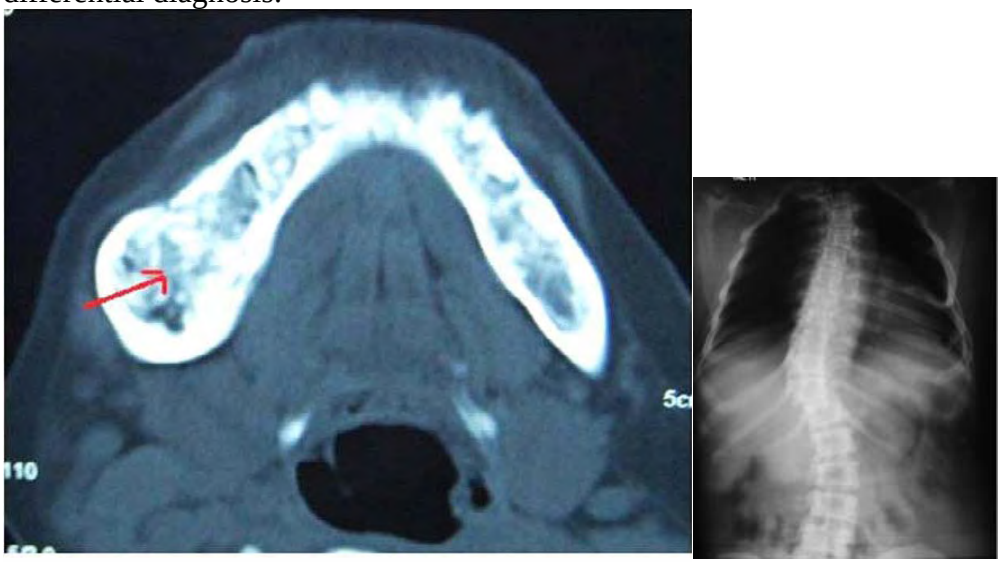
PMH

- The **patient's medical history** revealed the presence of **neurofibromatosis (NF)1** for 20 years, with neurofibromas all over the body and **caf'e-au-lait spots** present on the thigh and back with **regular borders** (coast of California). The patient reported that a history of NF1 ran in the family, and complained of generalized weakness, headache, fatigue, and pain in the lower limbs, joints, and back, which caused difficulty in walking. The patient was also a known **hypertensive** since 5 years of age, and was on medication. She had attained **menarche** at the age of 15 years.

X-ray

- Panoramic radiograph demonstrated a well-defined **mixed radio-opaque** and radiolucent lesion on the right body of the mandible, with ballooning of the inferior border. A computed tomography scan of the right body of the mandible showed an expansile mixed lesion (hypodense and hyperdense), with thinning of cortices and areas of hard and soft tissue attenuation, suggestive of FD, central

ossifying fibroma, and intraoral lesions of NF were considered under a differential diagnosis.



- The anteroposterior (AP) and lateral view of the spine showed the presence of **scoliosis**, and the lateral view showed thinning of the ribs. The AP and lateral view of the lower limbs showed an ill-defined mixed lytic-sclerotic lesion in the lower end of the right fibula with cortical thickening, suggestive of FD.
- A bone scan revealed an increased uptake of **pertechtenate** in the right body of the **mandible** and the lower end of the **right fibula**, suggestive of polyostotic FD. Dual energy X-ray absorptiometry scan showed decreased bone mineral density (BMD), with a T score of 2.7, suggestive of osteoporosis.

Table 1. Biochemical investigations

Laboratory values	Normal levels	Observed levels
Serum calcium (mg/dL)	9.00–10.40	8.40
Serum alkaline phosphatase (u/L)	40.00–136.00	151.00
Serum parathormone (mg/mL)	10.00–60.00	105.00
Serum phosphorous (mg/dL)	3.00–4.50	2.00
Serum creatinine (mg/dL)	0.60–1.20	0.60
Urinary phosphorous (mmol/L)	0.60–1.40	1.48
Urinary creatinine (mmol/L)	0.60–1.20	0.88
TmP/GFR (mmol/L)	0.15–0.24	0.08
1,25-dihydroxyvitamin D3 (pg/mL)	25.00–40.00	18.00
Thyroid stimulating hormone (Mu/L)	0.30–0.40	0.32
Thyroxin, total T4 (μg/dL)	4.50–10.90	8.50
Triiodothyronine, total T3 (ng/dL)	60.00–181.00	102.00
Growth hormone (ng/mL)	0.50–1.70	1.20
Cortisol, free (μg/24 h)	20.00–70.00	34.00

These levels were found to be suggestive of secondary hyperparathyroidismdue to vitamin D deficiency. Investigations for hypothyroidism, gigantism, and Cushing syndrome were performed to exclude McCune–Albright syndrome.

- Through an intraoral approach, curettage of the lesion was performed, followed by histopathological examination, which revealed trabeculae of woven bone with osteoblastic and osteoclastic activity without rimming of osteoblasts, suggestive of FD.
- A final diagnosis of NF1, polyostotic FD associated with osteoporosis, and secondary hyperparathyroidism due to vitamin D deficiency was established.

- The patient was prescribed **vitamin D** and **calcium** supplements. Follow up of the patient after 3 and 6 months showed remission in her symptoms.

Discussion：

1. Neurofibromatosis has been linked with FD of bone by various physicians, such as skin nodules, pigmented spots, and endocrine manifestations, which are often seen in FD.
2. **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**.
3. Rosenberg et al. reported a case series in which FD and NF were present in a **family** of nine. It has been speculated that this is more than a chance occurrence.
4. Additionally, the secondary manifestations of NF and FD, such as **osteoporosis**, **hyperparathyroidism**, and **osteomalacia**, are investigated in the present case.

Neurofibromatosis 1

1.Chromosome 17. Neurofibromin
2.Caf'e-au-lait spots
3. Bone: Short Osteoporotis Scoliosis
4. Hypertension
5. High level of PTH, low level of Vit. D3

Fibrous dysplasia

1. Guanine nucleotide stimulatory protein (GNAS1) gene
2. Fibroblast growth factor 23 increase
3. Renal phosphate wasting
4. McCune-Albright syndrome

Treatment with bisphosphonates is found to be beneficial in FD, resulting in rapid pain relief and normalization of bone turnover. Calcium and vitamin D supplements in patients with vitamin D deficiency are necessary to limit osteomalacia and hyperparathyroidism.

The management of NF1 is currently focused on genetic counseling and esthetic treatment of specific lesions, usually through surgery.

1	4. McCune-Albright syndrome 不包括以下何者 (A) polyostotic FD (B) Caf'e-au-lait spots (C) 性早熟 (D) Scoliosis
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題號	題目
2	Neurofibromatosis 1 常見的表徵 (A) Short stature (B) Osteoporotis (C) Acoustic schwannomas (D) Hypertension
答案(C)	出處：Journal of Investigative and Clinical Dentistry (2015), 6, 77–80