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Rare yet Resilient- Insights into Congenital Neurofibromatosis- Clinical Images

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CASE HISTORY:

A 42-year-old female patient presented to our hospital with a chief complaint of diffuse swelling on the right zygomatic region of the face, accompanied by multiple generalized cystic swellings distributed across both limbs, the abdomen, and the back. The largest of these swellings measured 2 cm x 1 cm. The swellings were asymptomatic, lacking pain and tenderness. The patient reported a history of similar swellings, for which she had undergone surgical excision twice previously. Despite these interventions, the swellings recurred, with the current recurrence being significantly larger than the previous ones. Notably, there was no history of pus discharge from any of the swellings, which have been present since birth. Additionally, the patient has a lipoma located over the left mid-back region, measuring 5 cm x 5cm x 3 cm, which has been present for the past four years. Her medical history is negative for hypertension, diabetes mellitus, tuberculosis, and thyroid disease. Surgical history includes excision of swellings one year prior.

KEYWORDS: Leopard syndrome, Lipoma, Neurofibromatosis, Parkes Weber Syndrome.

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Diagnosis:

Congenital Neurofibromatosis. The diagnosis was based on clinical signs and symptoms, which included multiple café au lait macules, intertriginous freckling, and multiple cutaneous neurofibromas.

Differential Diagnosis:

LEOPARD syndrome, Legius syndrome, Proteus syndrome, Macrodystrophia lipomatosa, Klippel-Trenaunay syndrome, Parkes Weber syndrome.

Clinical Image:

Image 1: Neurofibromatosis over face.

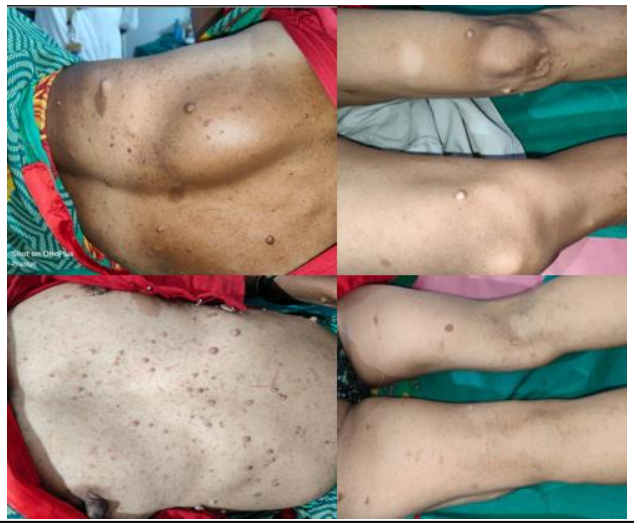

Image 2: Neurofibromatosis with lipoma over back and multiple neurofibroma over whole body.

DISCUSSION:

Neurofibromatoses are a group of hereditary disorders that result in tumor growth on nerve tissue. These tumors can develop in various parts of the nervous system, including the brain, spinal cord, and peripheral nerves. While most tumors associated with neurofibromatosis are benign, there is potential for malignant transformation.[1] Although symptoms are generally mild, neurofibromatosis can lead to significant complications such as hearing loss, learning disabilities, cardiovascular issues, blindness, and severe pain. [2]. Neurofibromatosis type 1 (NF1) is typically diagnosed in childhood, with signs often appearing by age ten, either at birth or shortly thereafter. The severity of symptoms can vary widely, but they are usually mild to moderate in most cases [3-4].

Clinical manifestations of NF1 include:

- **Café-au-lait spots**: These flat, light brown patches on the skin are benign and common in the general population. Having more than six café-au-lait spots is indicative of NF1. These spots are usually present at birth or appear within the first few years of life, with new lesions generally ceasing to develop after childhood [5].
- **Freckling**: Occurring in the groin or armpits by age three to five, these freckles are smaller than café-au-lait spots and typically appear in clusters within skin creases [6].
- **Neurofibromas**: These are pea-sized, soft lumps that can form on or under the skin. While these benign tumors often develop in or beneath the skin, they can also grow internally. Plexiform neurofibromas, which affect multiple

nerves, can cause disfigurement if located on the face. The number of neurofibromas may increase with age [7].

- **Skeletal abnormalities**: Bone deformities such as scoliosis or tibial dysplasia can arise from abnormal bone formation and reduced bone mineral density [8].
- **Optic gliomas**: Tumors of the optic nerve, typically presenting by age three. These tumors are rare in late childhood, adolescence, and adulthood [9].

Treatment options for abnormal tissue growth in neurofibromatosis include surgical excision of tumors, radiation therapy to destroy cancer cells and shrink tumors, and chemotherapy to eradicate tumors and prevent cancer spread. Chemotherapy is often used initially to reduce tumor size, making surgical removal more feasible [10-12].

Treatment Given:

The patient was prescribed Tab Kachnar Guggul 125 mg BD with lukewarm water after meals and Tab Arogyavardhini Vati 125 mg BD with lukewarm water after meals. The disease was not completely cured.

The purpose of publishing this clinical Image is to highlight its rarity, with a prevalence of 1 in 2500–3000. [13]

Consent of patient:

Consent was taken from the patient before starting the treatment protocol as well as prior to publication of the case details and data.

Conflict of interest: Author declares that there is no conflict of interest.

Guarantor*:* Corresponding author is guarantor of this article and its contents.

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