CLINICAL IMAGE

Neurofibromatosis

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ABSTRACT

A 52-year-old female presented with diffuse cutaneous involvement of Neurofibromatosis Type 1, with lesions distributed extensively over the face, scalp, neck, trunk, and both upper and lower extremities. The patient reported that several lesions had become tender over time, causing discomfort. Moreover, several lesions, owing to their size and anatomical location, were causing significant functional impairment, particularly with dressing and undressing, and were also compromising personal hygiene. In view of these symptoms, the patient sought surgical excision of the symptomatic and functionally limiting lesions.

Keywords: Neurofibromatosis, Neurofibromatosis, Schwannomatosis

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Neurofibromatosis is a group of inherited genetic disorders characterized by the development of tumours along nerves in the body. These tumours are usually benign but can sometimes undergo malignant transformation.

There are three main types:

1. Neurofibromatosis Type 1: is the most common type; causes skin spots (café-au-lait), small tumours on the skin, and sometimes bone or learning problems.
2. Neurofibromatosis Type 2: affects the brain and nerves, especially causing hearing loss due to tumours on the auditory nerves.
3. Schwannomatosis is rare; it causes multiple painful nerve tumours.

It is usually inherited (autosomal dominant), but can also occur spontaneously.

Currently, there is no "cure" that stops the tumours from forming. Management consists of:

- Managing the symptoms arising from the lesions.
- Monitoring: Regular "watch and wait" appointments with MRIs and vision/hearing tests.
- Surgery: To remove tumours causing pain, pressing on organs or affecting appearance.
- Drug Therapy: Specifically, for NF1, medications such as Selumetinib have been approved to help shrink plexiform neurofibromas in children.
- Genetic Counselling: Since NF is an autosomal dominant condition, there is a 50% chance that a parent will pass the gene to their child. However, about half of all cases result from a spontaneous mutation (neither parent had it).

Prognosis:

Most people with NF live productive, healthy lives. The key is early diagnosis and a multidisciplinary medical team (including neurologists, dermatologists, and geneticists) to stay ahead of any complications.

Conflict of Interest: None

Written Informed Consent: Taken

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