

A case of von recklinghausen disease

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ABSTRACT

A 67 years old man attended surgery OPD of Nepal Medical College and Teaching Hospital with complain of multiple painless swelling all around the body gradually increasing in numbers for last 15 years. Symptomatically some lumps were becoming large, covering skin of those swelling were sloughing with ulceration. Symptomatic lumps were excised. He was clinically diagnosed as Von Recklinghausen Disease. Histopathological report was Neurofibroma with non specific chronic inflammatory reaction.

Keywords: Neurofibromatosis, Painless swelling.

CASE REPORT

A 67 years old man attended surgery OPD with complain of multiple painless swelling all over the body. First swelling was noticed in the infraaxillary region at the age of 16 years. After that similar painless swelling started appearing in various parts of the body, which were gradually increasing in numbers since last 15 years. On examination his vitals were stable; Chest\ CVS-NAD.P/A-soft, non tender, no organomegaly. Multiple nodules of different sizes from 1x1 cm to 4x4 cm spread through out the body. Mostly these swelling were in the subcutaneous tissue and were pendulated. The nodules vary in consistency from very firm(even hard) to soft nodules.

A swelling over the left temporal region of size 10x5cm has lead to blockage of vision on that side. Since last 5 years he has been troubled with pain and bleeding from some of the lumps on several occasions and such troubling lumps were excised. In his family his elder daughter complains of similar type of painless swelling since the age of 21 years and which are gradually increasing in numbers.

Complete blood counts, ESR, X-ray chest all were normal. Histopathological Diagnosis of the lumps were Neurofibroma with non specific chronic inflammatory reaction.

DISCUSSION

Neurofibromatosis is an autosomal dominant disorder that affects the bone, the nervous system, soft tissues, and the skin. At least 8 different clinical phenotypes of neurofibromatosis have been identified and are linked to at least 2 genetic disorders. Neurofibromatosis has two forms Neurofibromatosis-1 and Neurofibromatosis-2. Neurofibromatosis type 1(NF-1), also called as Von Recklinghausen Disease, is a rare genetic disorder characterized by the development of multiple non cancerous(benign) tumors of nerves and skin (neurofibromas) an areas of abnormally decreased or increased coloration (hypo-or-hyper pigmentation) of the skin. It was discovered by Friedrich Von Recklinghausen. NF-1 gene is on the long arm of chromosome 17 and NF-2 gene on chromosome 22. The diagnosis Neurofibromatosis is on clinical basis and clinical manifestations increase over time, neurological problems and malignancy development may supervene.¹

NF-1 is caused by changes (mutations) of relatively larger gene on long arm (q) of chromosome 17 (17q11.2). The gene regulates the production of a protein known as Neurofibromin, which is thought to function as a tumor suppressor. In about 50 percent of the individuals with NF-1 results from spontaneous (sporadic) mutations of the gene that occur for unknown reasons. In others with the disorder, NF-1 is inherited as an autosomal dominant trait. Internationally worldwide NF-1 occurs in approximately 1 of 2500-3300 live births, regardless of race, sex, or ethnic, background. The carrier incidence at birth is 0.0004, and gene frequency is 0.0002. The incidence of NF-2 is 1 case per 50,000-120,000 population.²

This disease can involve various body system over time. Signs range from cutaneous manifestations

to profound disfigurement. Most individual who develop neurofibromatosis are not born with cafe au lait macules, these skin lesions develop during the first 3 years of life. Neurofibromatosis form in late adolescence may complain of cutaneous discoloration or disfigurement or more serious physical symptoms. Many individuals with neurofibromatosis have below average intelligence. Of patients with NF-1, 20.0-40.0% may have learning disabilities, while 5.0-10.0% may have mental retardation. The mortality rate is higher then that of the healthy population because of the increased potential for malignant transformation of diseased tissues and development of neurofibrosarcoma. Patient with NF-1 have an estimated 3.0-15.0% additional risk of malignant disease in their lifetime.³

Diagnostic criteria for NF-1(The diagnostic criteria are met if 2 or more of the features listed are present): 1. Six or more cafe au lait macules larger than 5 mm in greatest diameter in prepubertal individuals and those larger than 15 mm in greatest diameter in postpubertal individuals, 2. Two or more neurofibromas of any type or plexiform neurofibroma, 3. Optic glioma, 4. Flecking in the auxiliary or inguinal regions, 5. Two or more lisch nodules, 6. A distinctive osseous lesion, such as sphenoid dysplasia or thinning of the long bone cortex with or without pseudoarthrosis.7. A first degree relative with NF-1. Diagnostic criteria for NF-2 (The criteria are met if 1 or 2 is present):1.Bilateral masses of eighth cranial nerve seen with appropriate imaging techniques (eg: CT, MRI) 2.A first degree relative with NF-2.⁴

Currently mutation analysis using sophisticated genetic techniques which are 60.0-70.0% accurate in detecting these mutations, is available for NF-1 and NF-2.MRI of brain and spine may be helpful in patients with NF-1, specially if signs or symptoms suggests of Optic glioma a creteria for the diagnosis of NF-1 requires imaging for detection.⁵

Management of neurofibromatosis (NF-1and NF-2) is presently aimed at controlling symptoms with multidisplinary consultation. Surgery can help some NF-1 bone malformations and remove painful or disfiguring tumors. When neurofibromatosis increase in size or cause pain, malignant transformation should be suspected and excision or biopsy should perform. Acoustic neuroma and tumor that cause tinnitus and vertigo should be excised with great caution. Plastic surgeon may be included in the correction of deformities especially those of face. Psychological or psychiatric assessment may be necessary in monitoring language and learning disabilities. The skin tumors of Neurofibromatosis should not be excised unless they show evidence of malignancy or cosmetically objectionable. Response of benign lesions to radiotherapy is too weak to justify the risk of heavy exposure.⁵

The prognosis of neurofibromatosis is varied as is nature of the disease. In most cases, symptoms of NF-1 are mild, and patient live normal and productive lives. In some cases, however, NF-1 can be severely debilitating. In some case of NF-2, the damage to nearby vital structures, such as other cranial nerves and the brainstem, can be life threatening.⁶

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Fig. 1. Front side of body



Fig. 2. Multiple nodules the back side of the body



Fig. 3. Multiple nodules at the lateral side