Case Report:

Posterior Neck Gigantic Plexiform Neurofibromatosis

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Abstract

Neurofibromatosis (NF) is one of the most common genetic disorders. It is an autosomal dominant genetic disorder. It primarily affects the neural tissues. Neurofibromatosis type I (NF-1) also known as well as Recklinghausen's disease is the most common type. We present a case of a 62-year-old lady with NF-1. The disease was apparent since childhood with appearance of multiple hyper-pigmented skin macules. With time, more cutaneous lesions appeared and grew bigger all over the body surface. Because of huge neurofibromatosis over posterior neck, patient came for further treatment.

Keywords: Neurofibromatosis; neurofibromatosis type I; Recklinghausen's disease; plexiform; neurofibromatosis; neck

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Introduction

The diagnosis of NF-1 is made through clinical assessment including history and physical examination. These assessment findings are compared with standardized diagnostic criteria outlined by The National Institutes of Health (NIH).¹ The incidence neurofibromas over head and neck was account for 48%, while others like upper extremity (54%), lower extremity (31%) and most common manifestation was over the trunk (64%).² Plexiform type was found in 30% of patient with NF-1.³ Mass over posterior neck imposes challenges to a surgeon. As posterior neck contain vital structures such as spinal cord, occipital artery, subclavian artery, subclavian vein, cervical plexus, accessory nerve and vertebral body, it is always a challenging operation to a surgeon.

Case Summary

A 62-year-old lady presented with huge mass over posterior neck. She claimed the mass started since childhood and slowly growing with age. The mass disturbed her daily life activity. Other physical examination showed multiple café-au-

lait spots all over the body, freckles in the armpit and groin, multiple sessile or pedunculated masses over skin and scoliosis. She claimed had positive family history of neurofibromatosis type-1. Furthermore, she also complained of fever, poor oral intake, pain and foul-smelling discharge over the mass. Clinical examination revealed the mass was infected. Size of the mass 80 cm x 50 cm (Figure 1). The diagnosis was infected plexiform neurofibromatosis.

She was admitted for intravenous ampicillin/

Figure 1: Posterior neck huge plexiform NF measuring 80cmx50cm with evidence of superficial infection.

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sulbactam 1.5 gram 3 times daily for 5 days together with proper dressing using chlorhexidine and bactigras. Noted after 5 days in ward, patient developed productive cough, yellowish sputum and persistent spiking temperature. There were coarse crepitations on bilateral lungs auscultation hence was treated as nosocomial pneumonia. The antibiotic was changed to intravenous amoxicillin/clavulanate 1.2 gram 3 times daily.

Computed tomography (CT) scan of the neck and thorax revealed a huge cutaneous soft tissue pedicled mass arising from right posterior neck region measuring more than 40 cm in length and 20 cm in width (Figure 2). The stalk was about 5 cm x 2.5 cm. There was clear demarcation with surrounding structure. The underlying posterior neck muscle is normal. No calcification or fat content. No collection within visualized mass. There were dilated and tortuous vessels at right-sided of neck with present of dilated vessels within the mass arising from branches of right external carotid artery. The nasopharynx, oropharynx and laryngopharynx are normal. Visualized brain is normal. No mediastinum mass or lymph node enlargement. Bilateral pleural effusion with

Figure 2: CT scan (axial view) showed multiple dilated vessels within the mass.

adjacent lung collapse consolidation.

The patient agreed for excision of the mass. Removal of mass was done under general anesthesia. Intraoperatively, multiple dilated vessels was ligated with vicryl suture. Resection was made near to neck area using flap technique. The removed specimen weighted 6.7 kg and the estimated blood loss was 500 ml. Post-operation patient was kept in ICU another 2 days for observation before transferred out to general ward.

Figure 3: The mass weighted 6.79 kg

The total hospital stay was 12 days.

Patient was discharge well, there were no post operatively complications or morbidity. Histopathological examination revealed infected neurofibromatosis. Microscopically showed circumscribed unencapsulated lesion consist of bland spindle cells lesion with intervening wavy collagen bundles and focally myxoidstroma. There is focal ulcerated surface covered by collections of fibrinosuppurative material and neutrophils. No nuclear atypia, necrosis or increase mitosis. Immunostain with S-100 revealed diffuse positivity.

Discussion

Management of huge plexiform neurofibromatosis is usually surgery.⁴⁻⁶ Indications For Resection of plexiform neurofibromatosis include infection, cosmetic, intractable pain, neurological deficit, suspected malignant transformation, and progressive enlargement with compressive effects.⁷ In our case, the resection was done for infected mass, cosmetic and giant mass caused neck extension. Even with surgical excision, plexiform neurofibromatosis has a recurrence rate of 20%.⁸ Known risk of sequelae in NF is malignant transformation. In our case, the patient had the risk factor in view of long standing gigantic mass. Fortunately the histopathological examination was come back as infected NF. It was estimated that the risk of malignant transformation is around 5%.⁹ As any other soft tissue mass, MRI is the radiological investigation of choice to visualize the exact location, dimension and possible invasion of neurofibromatosis to the adjacent structures.¹⁰ As MRI was not available in our setting, we opted for CT scan. It was shown that the stalk of the mass was superficial to trapezius muscle and no invasion to adjacent structures.

The anatomical variation in this type of surgery is the great challenge besides the unclear tumor margin. Being in the posterior neck, the tumor may contain nerves and abundant blood vessels.

The feeding blood vessels were multiple, but major nerve was not identified. Resection of the tumor was successfully performed and the defect was covered using local skin flap. Post-operative the wound healed well.

Conclusion

Early surgical intervention may reduce the complication of plexiform NF. Cosmetically it

is easier to be performed if the lesion is small. Delay in treatment may cause rapid growing of mass, which can interfere daily life activity and develop risk of malignant transformation. Surgical excision and reconstruction with regular follow-up is an excellent treatment of choice for huge neurofibromas as in our case.

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