Congenital Neurofibromatosis in a Saudi Neonate who Presented with Neck Mass, Esophageal and Airway Obstruction

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ABSTRACT

We are presenting a case of a neonate presented with a neck mass, airway and esophageal obstruction, the tumor has a brain extension; treated with partial surgical excision; the pathological studies revealed plexiform Neurofibromatosis. The patient also has café au lait spots.

Key words:
Cervical plexiform neurofibromatosis, neck mass, esophageal and air way obstruction

INTRODUCTION

Neurofibromatosis type 1 (NF1) is one of the common autosomal dominant disorders with an estimated incidence of 1 in 4000.[1] Plexiform neurofibroma, however, is a very rare type of neurofibromatosis.

We are reporting a Saudi neonate who presented with a neck mass at birth with a serious compression effect on airways and esophagus, which proved to be plexiform neurofibromatosis.

CASE REPORT

A 15-day-old baby was referred to our center from a local hospital as a patient with right-sided neck mass noticed since birth. He was born to a 26-year-old mother with no antenatal problems and no family history of neurofibromatosis. The baby was intubated and ventilated due to respiratory distress at birth. On arrival to our hospital, he had noticeable right-sided firm mass involving the neck and lower face with undefined edge, irregular surface, and intact overlying skin [Figure 1]. Five *café au lait* spots were seen [Figure 2]. Two *café au lait* spots were in the left arm and measured 2 × 6 mm; two were in the back and measured 1 × 6 mm and 1.5 × 6 mm, respectively; and one was in the right leg and measured 6.5 × 1.5 mm. Magnetic resonance imaging (MRI) showed an extension of the mass into the skull base and the floor of the middle cranial fossa in the extra-axial space [Figure 3]. Incision biopsy obtained showed neural spindle cell neoplasm consistent with cellular neurofibroma with atypical feature. Microscopic description revealed spindle cell proliferation with wavy nuclei in myxoid background. There was mild nuclear polymorphism and focal moderate increase in cellularity. There were about 2 mitoses/10 HPF. There was no evidence of necrosis [Figure 4]. Immunohistochemistry showed the tumor cells were positive for S100 and Vimentin. They were focally positive for CD57 and CD56. They were negative for smooth muscle actin, muscle specific actin, Desmin, and CD68.

DNA studies were not done as they were not available in our center.

In the second stage, surgery was performed because of farther progression of the tumor, and the baby developed facial edema tracheostomy, gastrostomy tube and central line, as well as a partial decompression of the tumor. The surgical finding was extensive tumor occupying all of the right side of the neck, extending into the esophagus and involving the internal jugular vein with all of the lower cranial nerves. Patient tolerated the procedure well. There were no surgical complications. Few days later, we referred the patient back to the center in his hometown that referred him to us, making follow-up arrangements with our team.

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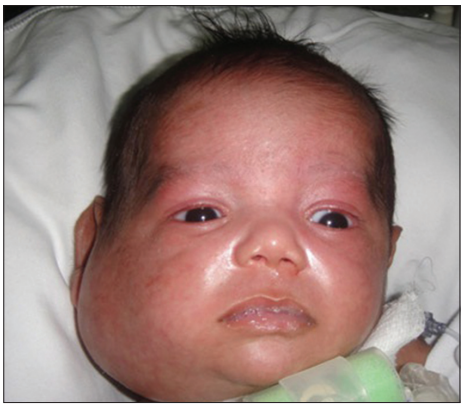

Figure 1: Patient before transfered back to the refered center showing the remnant of the tumor and the tracheostomy tube in place

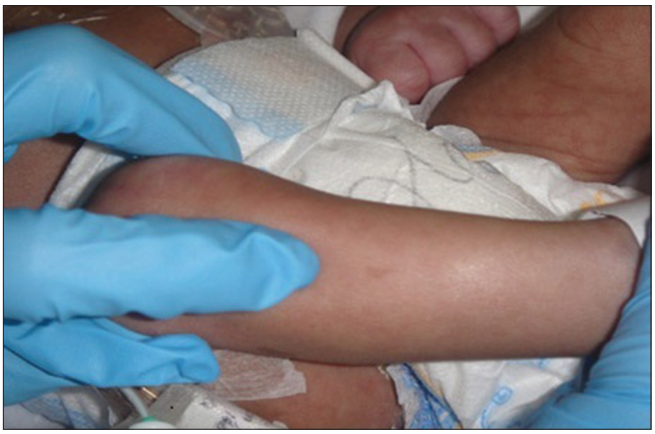

Figure 2: *Cafe au lait* spot

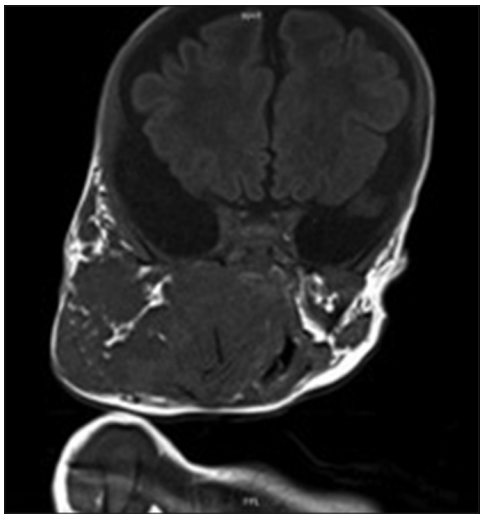

Figure 3: MRI showed an extension of the mass into the skull base and the floor of the middle cranial fosse in the extra-axial space

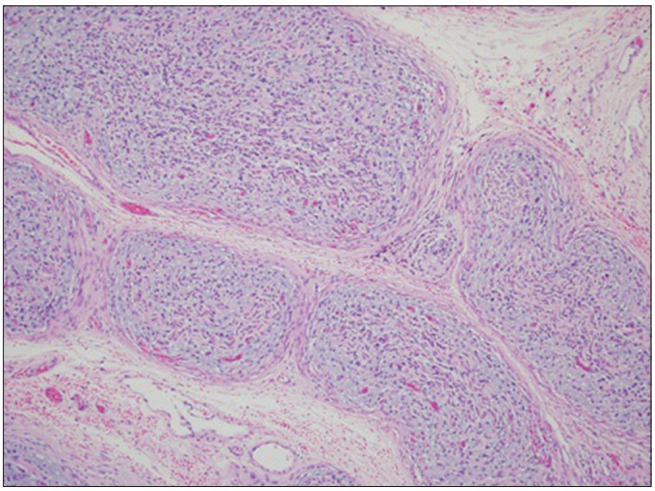

Figure 4: Plexiform neurofibroma expanding peripheral nerves, spindle cells in myxoid stroma dissecting through nerve fibers and contained by perineurium

DISCUSSION

The common presenting findings in congenital neurofibromatosis are hydrops fetalis, macrocephaly, *café au lait* maculae, skin nodules, buphthalmos, enlarging, proptotic glaucomatous eye, and rarely brain tumors.[1]

Plexiform neurofibromas may present at birth or become apparent during the first year of life in 30% of patients diagnosed with neurofibromatosis type 1.[2] Cranio-orbito-temporal lesions exist in about 1% of the cases;[3] however, in our patient, it showed an extreme picture of the disease with brain extension. Plexiform neurofibromas often cause disfigurement, progressive neurologic deficits, unremitting pain, compression and infiltration of vital structures, and thus represent a source of major morbidity and mortality in neurofibromatosis.[4] In our patient, it caused serious airway obstruction, which required early intubation followed by tracheostomy, and esophageal compression, which required orogastric tube for feeding. Subsequently gastrostomy tube.

These tumors do not regress spontaneously, and in many patients their growth is relentless.[5]

The management of plexiform neurofibromas is limited to surgical resection, but complete tumor removal is rarely possible due to the size, location, and infiltrating nature of the tumor.[3] It was successfully done in our patient.

CONCLUSION

Plexiform neurofibromatosis rarely presents with neck mass at birth, brain extension, and serious airway and esophageal obstruction.

We would like to bring to the attention that a neck mass with airway obstruction should make the clinician alert to look for intraprenchymal brain extension and diagnostic features of neurofibromatosis such as *café au lait* spots.

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