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Prenatal Diagnosis of Foetal Cervical Neuroblastoma: Report of a Case and Review of the Literature

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Abstract

Neuroblastoma is the most common extra cranial solid tumour in childhood and in infancy, but it is a rare fetal tumour, especially on neck region. Approximately, two thirds of the cases arise from adrenal gland and presentation of other localizations is extremely rare. Here, we report a case of cervical Neuroblastoma suspected at 20th gestational week. This is the fourth case detected on prenatal ultrasound on neck region and these cases that is identified in the second trimester fetal anatomy screening.

Keywords: Neuroblastoma; Adrenal gland; Ultrasound

Introduction

Neuroblastoma is a sarcoma arising from pluripotent sympathetic cells called neuroblast. It is the most common malignant tumor diagnosed in the neonatal period and the prognosis of the disease depends on the tissue of origin, age at diagnosis, stage of disease, cytogenetic abnormalities, and associated congenital malformations [1,2]. Differential diagnosis of fetal neck tumours is quite difficult. A widespectrum of diseases may be possible reasons like lymphangioma, teratoma, rhabdomyoma, sarcoma, haemangioma, neuroblastoma, goitre, and more [3]. The exact prenatal diagnosis of this tumor is of great importance for both maternal and fetal well-being as well as neonatal outcome. Here, we present a fetal cervical neuroblastoma detected on second trimester fetal anatomical scan at 20 weeks gestation.

Case

A 30-year-old pregnant woman (gravida 3, para 2) with no remarkable history of accompanying disease was referred to our clinic because of a fetal neck mass, identified on second trimester anatomical scan. The patient was not taking any medication other than daily vitamin pills. She did not have history of exposure to radiation or any other teratogen and there was no family history of congenital malformations. The patient was evaluated with level 2 fetal sonogram again. Level 2 sonogram, which was obtained with a Voluson E8, revealed a left sided lateral neck mass sized 40.22x41.51mm. The mass was heterogenous and solid in nature with few cystic components and it was extending from the level of clavicula to brainstem (Figure 1). The tumor was just near the trachea but was not obstructing it yet. In an effort to obtain more information and to confirm the diagnosis fetal MRI was performed at the day of sonography. The result was supporting our sonographic findings (Figure 2). We explained the family maternal and foetal effects of the situation and offered them termination of pregnancy. But they refused this option and after their refusal we obtained informed consent about the scenario waiting them. At the gestational age of 34 weeks and 5 days the patient who was in the active stage of labor, again referred to our clinic. She delivered a girl weighing 2500 gr by caesarean section because of the fetal neck mass and previous cesarean sections. After the delivery the baby did not have spontanous respiration, because of the neck mass intubation could not be done promptly. Emergency tracheostomiy was made by otolaryngology specialist. But at the 12 th hour of life, she died because of respiratory insuffiency and subsequent multiple organ failure.

"Figure 3"shows the baby during the effort for opening the airway.

The diagnosis was confirmed by histopathological examination of the biopsy specimen taken from the mass, in post-mortem period. Thebiopsy specimen was stained with NSE (Neuron Specific Enolase). Immunohistochemical stainings for keratin, smooth muscle actin, vimentin, desmin were negative. And also, N-myc amplification was negative (Figure 4).

Discussion

Neuroblastoma, which usually presents with an adrenal mass, is an embryonal cancer consisting of malignant neuroblasts that can be found in many locations within the body [4]. Neonatal incidence of neuroblastoma has been reported between 58- 61 per million and has a male dominance [5,6]. With the increasing prevalance of routine fetal anatomical scans, prenatal diagnosis rates of this entity started to increase. But, still there are just a few number of case reports. Impaired folate metabolism, folate deficiency and history of fetal loss are shown as the risk factors for fetal neuroblastoma [7].

Since most of the cases arise from adrenal origin, extra adrenal neuroblastomas can be unnoticed and rarely diagnosed during pregnancy. Especially, neuroblastoma on neck region is extremely rare. Gadwood and Reynes were the first to diagnose a neuroblastoma on fetal neck region at 1983 [8]. With the limited capabilities of devices that was just an enormous prenatal diagnosis. Most of the prenatal diagnosis of extra adrenal neuroblastomas was madeduring the third trimester. Because, they are likely too small to be detected earlier. Gorincour et al. [9] reported the second case of fetal neuroblastoma on the neck at 2003. As well as the first case, this one was detected in the third trimester, too. At first, they assumed that it was a rhabdomyoma. But, biopsy of the cervical mass revealed a neuroblastoma [9].

Güzelmansur et al., reported the third neuroblastoma case on neck region at 2011. The difference of this case was the extension of the mass to fetal brain and the diagnosis at 20 weeks and 5 days of gestational age. Our case is second trimester diagnosis and extension of the mass to fetal brain. The only difference between the two cases is the time of delivery. Because of the refusal of the family, our case gave birth at the third trimester.

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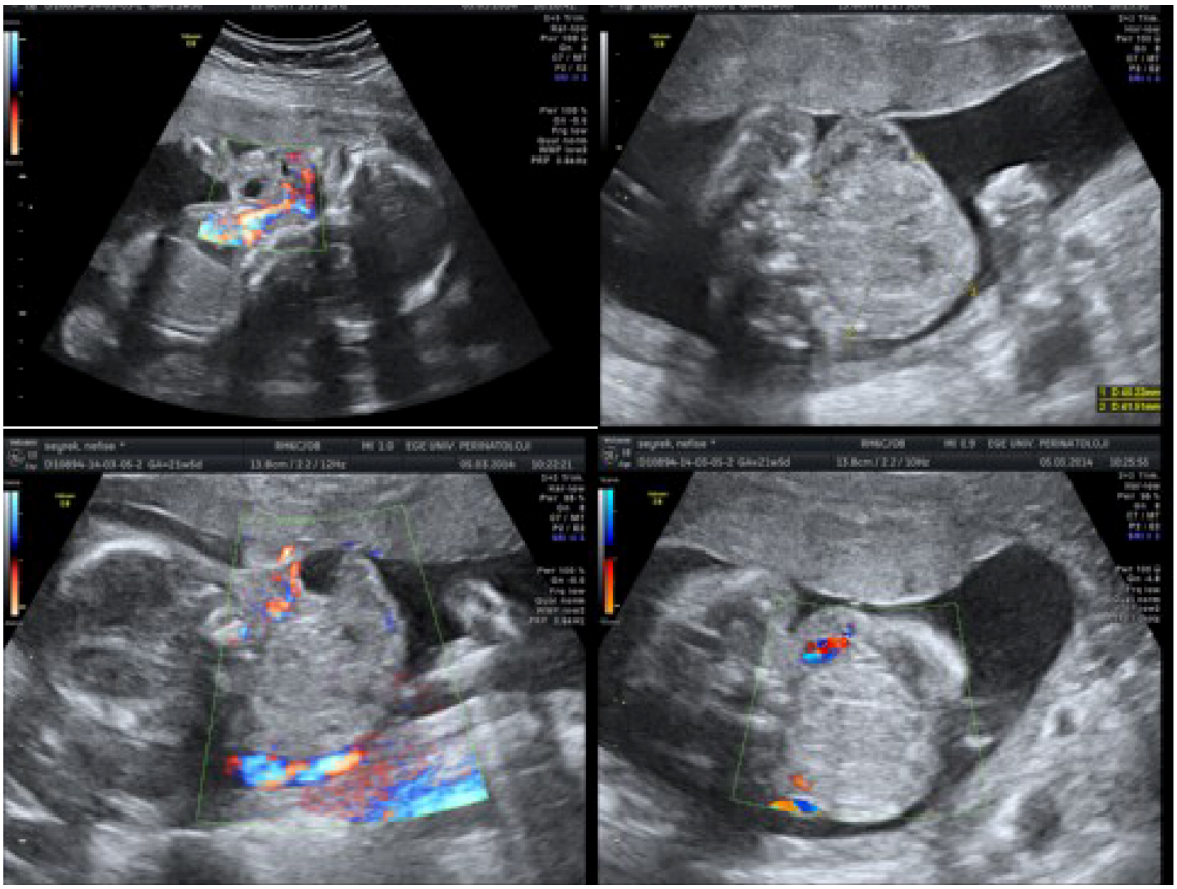

Figure 1: İntrauterin sonographig findings the mass was heterogenous and solid in nature with few cystic components and it was extending from the level of clavicula to brainstem.

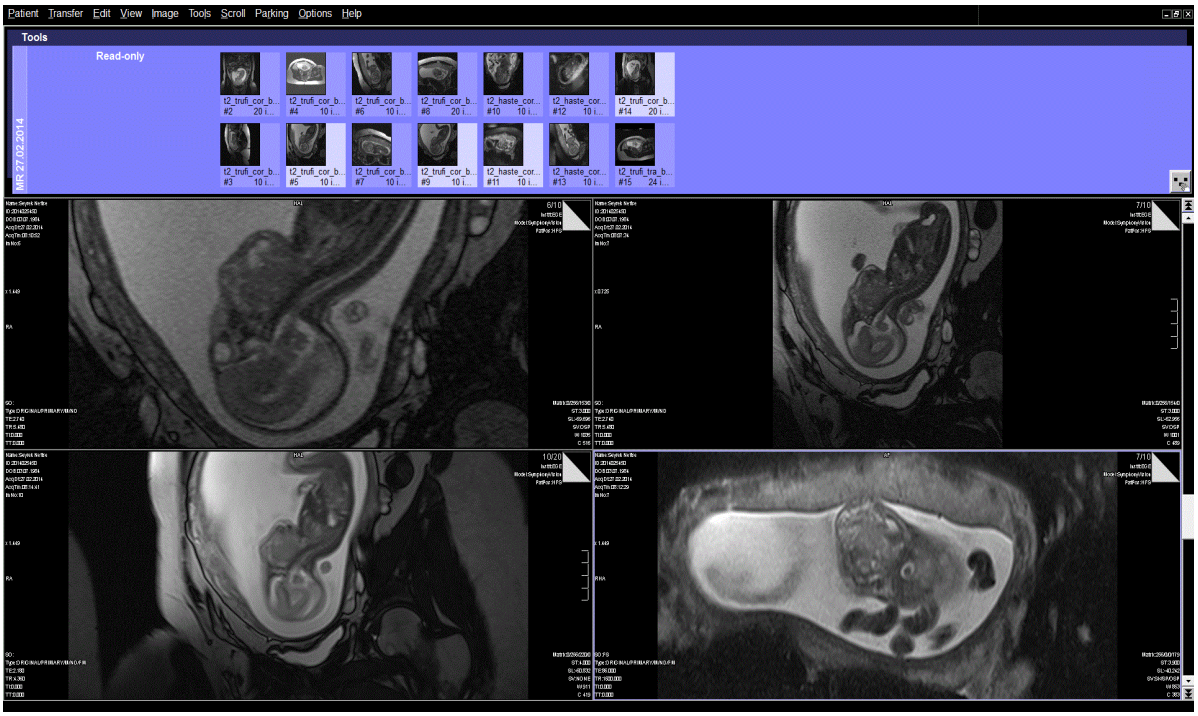

Figure 2: İntrauterin fetal MRI.

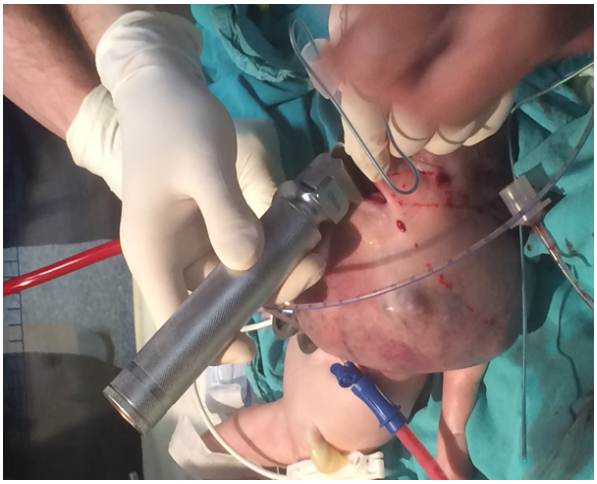

Figure 3: Shows the baby during the effort for opening the airway.

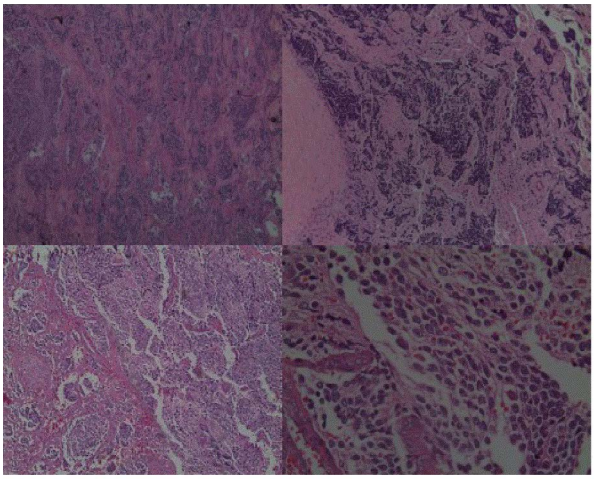

Figure 4: Histopathological examination of the biopsy specimen taken from the mass.

As we mentioned before, the origin of a fetal mass can be at any point on a wide spectrum. Tumors in cystic nature are generally lymphangiomas and higromas, while the tumors in solid nature are generally teratomas. But also, diagnosis like sarcomas, hamartomas, fetal goiter and rarely tuberous sclerosis may be possible. D. F´enart et al. [10] was the first to define a neuroblastom aprenatally, at 1983 [10]. They described the lesion as "cystic and around 50% having solid components and also calcifications may be present". The detection of has been associated with improved survivals how in previous tumor necrosis, whereas the presence of solid components are associated with metastases. Especially, in cases in which sonographic findings are unclear, fetal MRI is an important adjunct to prenatal sonography [11]. Owing to its multiplanar imaging capability and ability to differentiate blood products for mother tissues, it is very useful in making the exact diagnosis and showing the metastases. As a conclusion, however it is a rare disease in prenatal period, neuroblastoma should be in the list of differential diagnosis of a fetal neck masses. In view of its possible adverse effects on both maternal and fetal health, termination of pregnancy should be considered as an option.

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