Yeditepe Medical Journal 2013;7(28): 705-710

ASSOCIATION OF RHABDOMYOMA, TUBEROUS SCLEROSIS AND CONGENITAL ADRENAL HYPERPLASIA: AN UNUSUAL NEWBORN CASE

Case Report

ALIŞILMADIK BİR YENİDOĞAN OLGUSU: RABDOMYOM, TUBEROZ SKLEROZ VE KONJENİTAL ADRENAL HİPERPLAZİ BİRLİKTELİĞİ

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ABSTRACT

Tuberous sclerosis is a neurocutaneous syndrome involves skin, brain, kidneys, heart and eyes primarly. Cardiac rhabdomyomas accompany to tuberous sclerosis frequently. Congenital adrenal hyperplasia is group of autosomal disorders of adrenal cortex characterized with enzyme deficiency at cortisol biosynthesis. There is only one case with tuberous sclerosis and congenital adrenal hyperplasia reported in literature.

We report a case with intracardiac masses which were detected in the left ventricle with prenatal and postnatal echocardiography. Tuberous sclerosis with subependymal nodules and cortical tubers was detected at postnatal magnetic resonance imaging. Also congenital adrenal hyperplasia was determined at the infant which had areolar and scrotal hyperpigmentation.

Key words: *Rhabdomyoma; tuberous sclerosis; congenital adrenal hyperplasia; newborn.*

ÖZET

Tuberoz skleroz, öncelikle cilt, beyin, böbrekler, kalp ve gözleri etkileyen bir nörokutanöz sendromdur. Kardiyak rabdomyomlar tuberoz skleroza eşlik edebilirler. Konjenital adrenal hiperplazi, adrenal korteksin kortizol sentezindeki enzim defekti ile karakterize otozomal hastalıklar grubudur. Literatürde, tuberoz skleroz ile konjenital adrenal hiperplazi

birlikteliği olan sadece bir olgu bildirilmiştir.

Prenatal ve postnatal dönemde ekokardiografi ile intrakardiak sol ventrükülde kitleler saptanan bir olgu sunulmuştur. Olguda postnatal dönemde yapılan manyetik rezonans görüntülemede subependimal nodullerin ve kortikal tuberlerin olduğu tuberoz skleroz tanısı konuldu. Ayrıca areolar ve skrotal hiperpigmentasyonu olan olguda konjenital adrenal hiperplazi saptandı.

Anahtar kelimeler: *Rabdomyom; tuberoz skleroz; konjenital adrenal hiperplazi; yenidoğan.*

INTRODUCTION

Tuberous sclerosis is a neurocutaneous syndrome characterized with tumoral and nontumoral proliferations, anomalies, usually ocurring in the brain, skin, eyes, kidneys, heart and lungs. Tuberous sclerosis is an autosomal dominant disorder with cerebral cortical anomalies, subependimal tumors, seizure, mental deficiency, renal anjiomyolipoms and cardiak rhabdomyoma (1,2). Cardiac rhabdomyomas have been seen 43-60% with tuberous sclerosis. Cardiac rhabdomyomas are the most common cardiac tumors in childhood (3) and are usually diagnosed after 32nd weeks with ultrasound prenataly. So that tuberous sclerosis shold be connoted when cardiac rhabdomyoma is confirmed (3).

Congenital adrenal hyperplasia is a metabolic and endocrin disorder which inherites autosomal recessively and causes insuffciency at enzymes neccessary for adrenal steroidogenesis. Deficiency of 21-hydroxylase is the most cause of congenital adrenal hyperplasis. More than 90% of congenital adrenal hyperplasias are caused by 21-hydroxylase deficiency (4).

Only one case with congenital adrenal hyperplasia and tuberous sclerosis association was reported in the literature (5). We reported a case had tuberous sclerosis with rhabdomyoma, subependymal nodules, cortical tubers and congenital adrenal hyperplasia with hyperpigmentation, hyperkalemia, hyponatremia and high level 17 OH progesterone and progesterone.

CASE REPORT

We reported a case that intracardiac three masses were detected at him with prenatal ultrasound and that was hospitalized in neonatology unit posnataly (**Figure 1**).

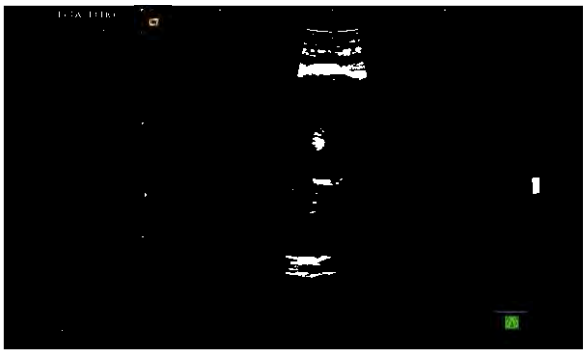

Figure 1:*Intracardiac masses at prenatal echocardiography.*

This was the eighth pregnancy of 37 years old mother. She had two babies who were died when 1 and 6 mounthly because of pulmonary failure and ambigius genitalia. She had also 3 abortus and have 2 healthy children. There was a consanguinity between mother and father. Intracardiac three masses were determined in in left ventricle at routine fetal echocardiography which is performed at 37th weeks of gestation.

The male infant was born with elective caesarean section at 38-39th weeks of gestation. The birth weight was 3800 g (25-50th percentile), the length was 51 cm (90th percentile) and the head circumstance was 35 cm (50-75th percentile). His arterial blood pressure was 69/45 mmHg and arterial oxygen saturation was 99 %. Hyperpigmentation was detected at areola and scrotum.

Cardiac oscultation and the other examination findings were normal (**Figures 2 and 3**).

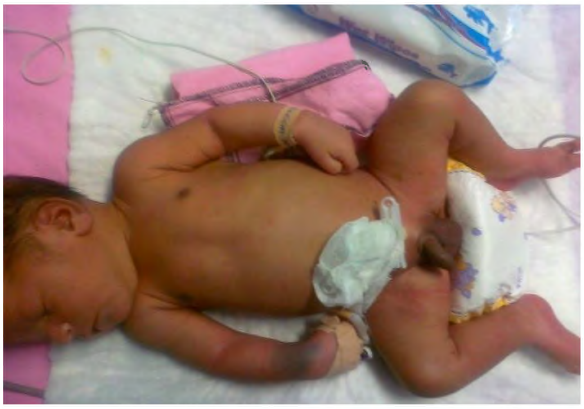

Figure 2. *General appearance of the patient*

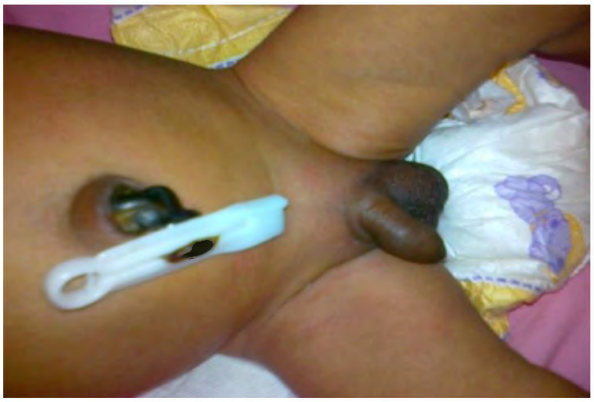

Figure 3. *Scrotal hyperpigmentation.*

Hemoglobin was 17.1 g/dl, hemotocrit was 47.6 %, white blood cell was 16600/mm^3, platelet was 334000/mm3 at CBC. 68 % PNL, 32 % lymphocyte were detected at peripheral smear. Renal and liver function tests, cardiac enzymes were normal. Blood sodium was 136 mEq/L, potassium was 4,6 mEq/L and glucose was 108 mg/dl. Also electrocardiogram (**Figure 5**) and telecardiography were normal (**Figure 6).**

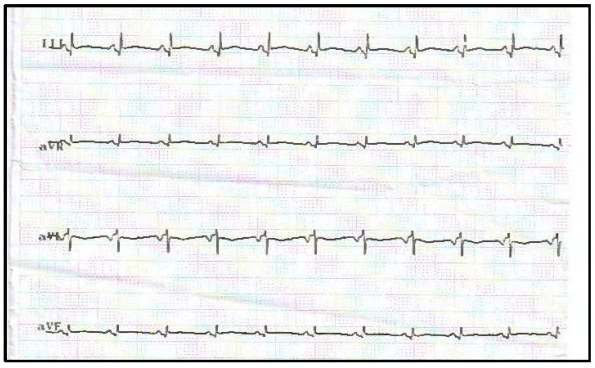

Figure 4. *Electrocardiogram .*

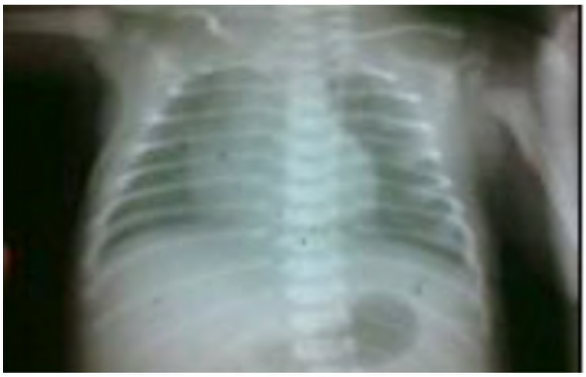

Figure 5. *Telecardiography*

Two masses were determined at echocardiography and they were described as rhabdomyoma. The dimensions of the masses were 15*10 mm and 16*10 mm. They protruded into the left ventricul. (**Figures 6 and 7**). Therefore tuberous sclerosis was investigated. Cranial magnetic resonance imaging showed subependymal nodules at the lateral ventricle side wall and cortical tubers at the frontal lobe . These findings were interpreted as tuberous sclerosis. (**Figures 8 and 9**). Renal ultrasound and retinal anjioscopy were normal.

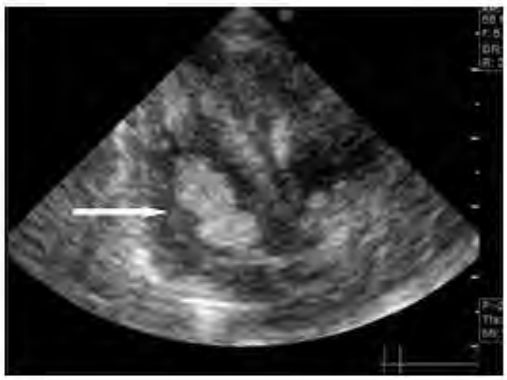

Figure 6. *Masses at echocardiography*

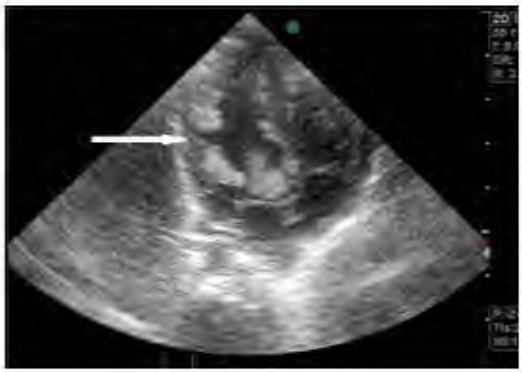

Figure 7. *Masses at echocardiography.*

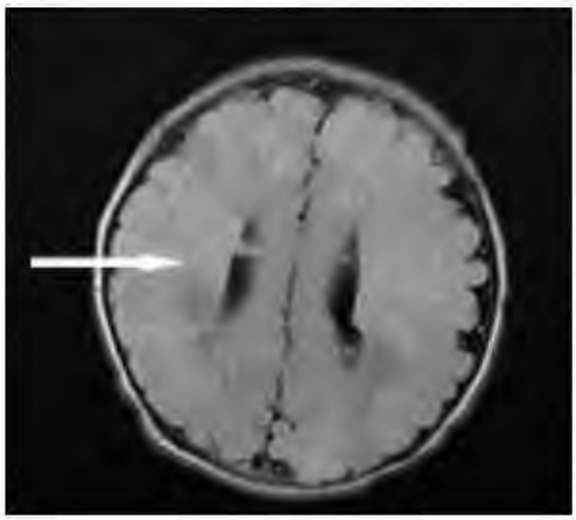

***Figure 8*.** *Subependymal nodules at MRI*

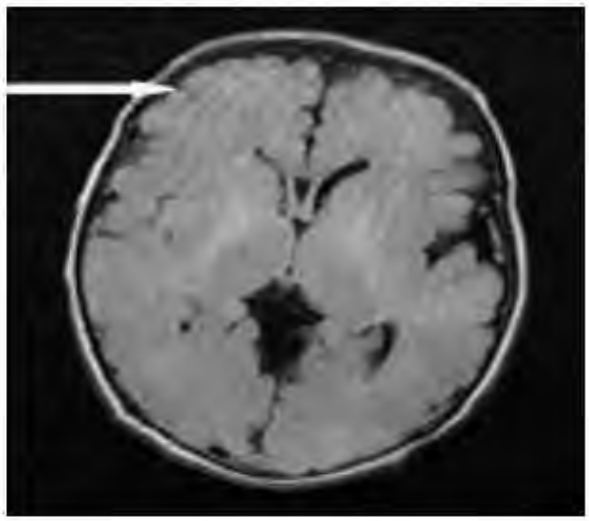

***Figure 9*.** *Cortical tubers at MRI*.

During the follow up, rhythm anomalies did not occur and the heart rate was 100-140 bpm. Cardiac rhabdomyomas did not cause obstruction and arrhythmia so surgical and antiarrhythmic medical treatment were not applied. We followed intracardiac masses weekly by echocardiography because of spontaneous decreasing.

Congenital adrenal hyperplasia was investigated because of areolar and scrotal hyperpigmentation. Also there was a sister died with ambigius genitalia and was a consanguinity between mother and father. Progesterone was > 40.80 ng/ml (0.10-0.84), 17-OH progesterone was 40.34 ng/ml (0.5-2.4), ACTH was 88.5 pg/ml (5-46) and DHEAS was 62 mcg/dl (80-560) at laboratory study. At the ninth day of follow up, blood sodium was 127 mEq/L, potassium was 7.2 mEq/L and glucose was 110 mg/dl. Congenital adrenal hyperplasia was thought and hydrocortisone (20 mg/m2), fludrocortisone (0.05 mg/day) and salt (2 g/day) treatments were started.

DISCUSSION

Tuberous sclerosis is a disorder involving multiple systems mainly causes that mental deficiency, seizure and adenoma sebaseum. Tuberous sclerosis is

an autosomal recessive inherited desease. Rhabdomyoma is the most common cardiac tumor in childhood and usually accompanies tuberous sclerosis. Rhabdomyoma is important for early diagnose at tuberous sclerosis (1,3,6,7).

Cardiac rhabdomyomas are nodular masses, usually localized in the ventricules or less commonly in the atrium (8). Their size can vary from a few millimetres to massive tumours able to obstruct outflow of one or both ventricules. They may be single or multiple (90% of cases) and tend towards spontaneous regression. As a consequence they may be totally asymptomatic and incidentally discovered on echocardiograms or may also cause cardiac dysfunction requiring medical or surgical interventions. Life threatening conditions asssociated with cardiac rhabdomyoma due to arrhythmias, cardiac failure or obstruction are rarely reported in the neonatal period (8-12). We reported a case with multiple cardiac rhabdomyomas in left ventricle which didn't cause symptomatic conditions.

When cardiac rhamdomyomas are diagnosed prenatally, coordinated delivery planning before the occurence of haemodynamic complications can affect fetal and neonatal outcomes. In the case of life threatening situations, the neonatal outcomes is more influenced by obstructive rather than by dysrhythmic forms of cardiac rhabdomyomas if appropriate antiarrhythmic treatment is begun promptly. If acute obstruction to blood flow persists, surgical excision may be lifesaving (9,13). In this case, cardiac rhabdomyomas didn't cause haemodynamic complication, obstruction and arrhythmia. So that we didn't apply any surgical or medical treatment. We followed up cardiac rhabdomyomas by echocardiograms.

Salt wasting form of congenital adrenal hyperplasia is one of the most common cause of adrenal cortex insufficiency. Congenital adrenal hyperplasia results because of lossing activity of the enzymes necessary for adrenal steroidogenesis. The disease is characterized with decresed cortisol, aldosterone and androgen production or excessive cortisol, aldosterone and androjen production. Depending on the deficient enzymatic step, there may be signs, symptoms and laboratory findings of mineralocorticoid deficiency or excess; incomplete virilization or sexual infantilisim in affected females. Cortisol and aldosterone deficiency caused salt wasting form of the disease. Cortisol deficiency increased secretion of ACTH and hyperpigmentation is caused by ACTH [14,15]. This case had hyperpigmentation, and hyponatremia, hyperkalemia of salt wasting signs. The genital organs were normal and hypotension, hypoglycemia signs didn't occur in our case. We evaulated this case as salt wasting form of congenital adrenal hyperplasia because of these signs and symptoms. More than 90% of congenital adrenal hyperplasia cases are caused by 21-hydroxylase deficiency (4,16). 21-hydroxylase deficiency occurs one per 14200 births (17). Approximately 75% of affected infants have the salt losing form, whereas 25% have the simple virilizing form of the disorder. Patients with salt wasting disease have typical laboratory findings associated with cortisol and aldosterone deficiency, which is including hyperkalemia, hyponatremia, and hypoglycemia, but abnormalities can take 1-2 weeks or longer to develop after birth (4). Laboratory findings associated with aldosteron deficiency including hyponatremia, hyperkalemia were developed on 9th days after birth at our case. Blood levels of 17-hydroxyprogesterone and progesterone were eleved at our case. These signs and symptoms were resolved after hydrocortisone and fludrocortisone treatment.

Only one case with association of tuberous sclerosis and congenital adrenal hyperplasia reported in the literature [5]. Our case is the second report in the literature. Consequently rhabdomyoma

should be connoted when intracardiac masses are determined at prenatal ultrasound and then tuberous sclerosis should be investigated at postnatal period. Also congenital adrenal hyperplasia shold be investigated when there are family story and hyperpigmentation because of autosomal recessive inheritance.

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