

# INTRACARDIAC TUMOUR AND BRAIN LESIONS IN TUBEROUS SCLEROSIS

## A case report of antenatal diagnosis by ultrasonography

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### Abstract

A case of antenatal tuberous sclerosis was diagnosed by ultrasonography. Intracardiac tumour (highly suspected rhabdomyoma) with transitory heart failure and multiple brain lesions were observed. After delivery, echocardiography, spiral CT and MR imaging diagnosis of tuberous sclerosis was confirmed by typical skin lesions (depigmented macules) and development of seizures.

*Key words:* Tuberous sclerosis; heart, neoplasms; brain lesions; ultrasonography.

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Tuberous sclerosis (Bourneville Pringle disease) is an autosomal dominant congenital disease due to gene locus in chromosomes 9 and 16. The disorder has widespread potential for hamartomatous growths in multiple organ systems (6, 9). The clinical manifestations include the following: 1) Skin lesions: fibrous-angiomas (adenoma sebaceum), pigmented naevi, leucoderma (depigmented macules). 2) Neurological: seizures, mental deficiency, developmental deficiency. 3) Cardiovascular: hypertension, aneurysms, cardiac rhabdomyoma, arrhythmias, heart failure. 4) Renal: angiomyolipomas, renal failure, renal haemorrhage. 5) Respiratory system: cor pulmonale, pneumothorax. 6) Retinal: phacomas.

The incidence of cardiac tumours in fetuses or neonates is 0.1% in paediatric cardiac units and 0.0017–0.25% in *post-mortem* data (5). Rhabdomyoma is the most common intracardiac tumour in this group of patients and in 51–86% associated

with tuberous sclerosis (4, 5). We present a case of this rare congenital anomaly, mostly described in paediatric and cardiology literature. To our knowledge, antenatally diagnosed intracardiac tumour with foetal heart failure and subsequent treatment is exceptional.

### Case Report

In 1998, a case of cardiac tumour was diagnosed during a routinely performed antenatal ultrasonographic (US) examination at the 31st week of pregnancy in a healthy woman aged 41 years. The child's father was also healthy.

US revealed a cardiac tumour approximately 31×22 mm in size located in the right atrium and the right ventricle with involvement of the interventricular septum (Fig. 1a). Subsequent antenatal examinations showed increasing cardiac tumour size and slight hydrops was noticed. In the foetal

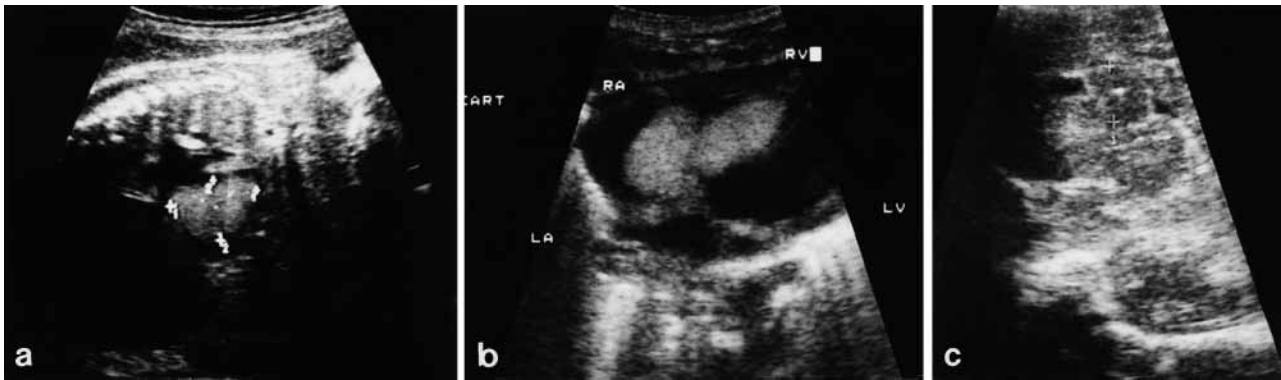


Fig. 1. a) Antenatally performed US at 31st gestation week showing intracardiac tumour (31×22 mm). b) Postnatal US of the heart in a four-chamber view. Rhabdomyoma occupying the right atrium and the right ventricle with involvement of the interventricular septum. (RA right atrium, RV right ventricle). c) Postnatal US of the brain showing a tuberous lesion in the right hemisphere close to the corpus callosum indenting the anterior horn of the right lateral ventricle.

peritoneal cavity, slight amounts of free fluid and dilated vena cava inferior and hepatic veins were present as evidence of heart failure. Arrhythmias were also revealed. A few intracerebral lesions were diagnosed, including one 13×11 mm lesion located periventricularly on the right side and subependymal in the left ventricle's anterior horn. Tuberous sclerosis was strongly suggested.

Delivery was performed by Cesarean section be-

cause of antepartum vaginal haemorrhage at 36 weeks.

Echocardiography confirmed prenatally diagnosed intracardiac tumour, which was well seen in a four-chamber view (Fig. 1b) occupying most of the ventricular cavity and 40×22 mm in size.

US of the brain revealed one 16×11-mm solid mass in the right hemisphere and small subependymal lesions in the left lateral ventricle (Fig. 1c).

CT demonstrated slight hyperdense lesions adjacent to the corpus callosum and the anterior horn of the right lateral ventricle, and three smaller lesions subependymally in the left lateral ventricle. Abnormal gyri could be seen at the surface of the brain temporally and parietally on the right side (Fig. 2). Spiral CT of the chest and heart



Fig. 2. CT of the brain showing the right hemispheric lesion (15×16 mm) lateral to the anterior horn of the right ventricle. There are small subependymal lesions in the left lateral ventricle. Brain gyri located frontotemporally on the right side show abnormal size and shape due to tuberous sclerosis.



Fig. 3. Spiral CT of the chest in the axial plane delineates a contrast medium filling defect due to intracardiac tumour occupying a large part of the right atrium and ventricle.

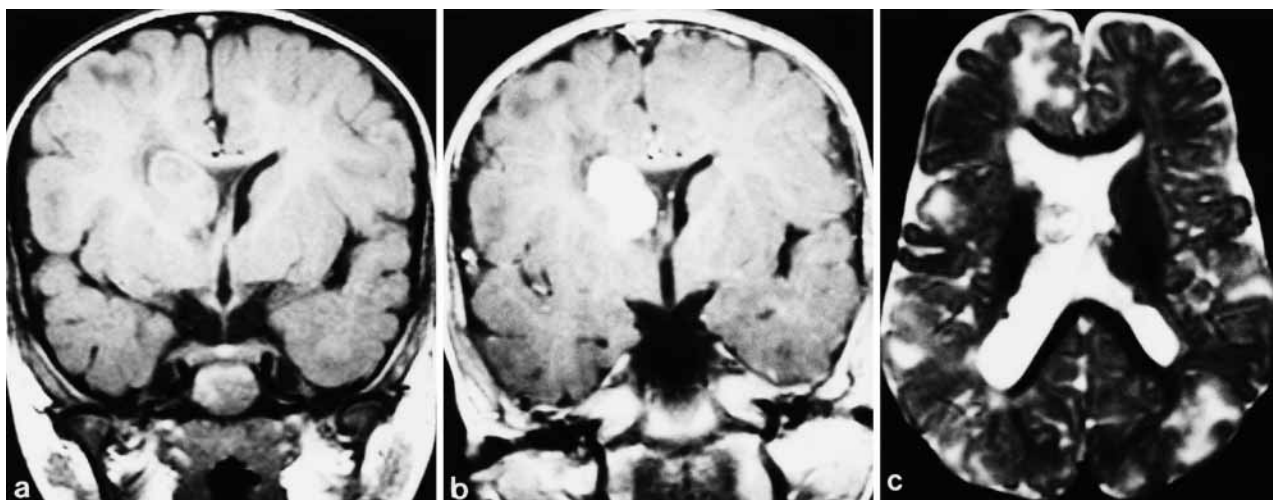


Fig. 4. T1-weighted image in the coronal plane (a) shows an approximately 2×1 cm mass located subependymally and bulging into the right frontal horn with (b) intense homogeneous contrast enhancement. It extends down to the level of the foramen of Monro. It has the appearance of a giant cell astrocytoma. c) Multiple cortical and subcortical high signal intensity lesions are detected in the cerebrum on the T2-weighted image. The subependymal giant cell astrocytoma is bulging into the right lateral ventricle.

delineated the topography of the tumour in axial, coronal and sagittal planes (Fig. 3).

The patient had two cutaneous lesions (ash-leave-shaped depigmented macules) at the body trunk, characteristic of tuberous sclerosis.

At the age of 19 days, the child was transferred to Great Ormond Street Hospital for Children in London for further management. Cardiosurgical intervention was not undertaken. The tumour did not grow and there was no sign of heart failure. During observation in London, the intracardiac tumour had slightly diminished in size. At the age of 10 months, the patient developed seizures, which are now controlled by pharmacological treatment. Otherwise good development and good weight and length increase were observed. At that time MR examination was performed without and with i.v. contrast injection, which showed typical appearance of tuberous sclerosis with right-sided subependymal giant cell astrocytoma (Fig. 4).

### Discussion

This case presented the typical characteristics described in tuberous sclerosis: lesions in brain and intracardiac tumour (rhabdomyoma), and cutaneous location as well, with subsequent seizures fulfilling the diagnostic criteria. An intracardiac tumour in the antenatal period is mostly due to rhabdomyoma, which together with brain lesions strongly suggests tuberous sclerosis (2, 3, 5). The diagnosis of tuberous sclerosis in our case was confirmed using criteria adapted by multicentre

studies by HOLLEY et al. (5), which are as follows: 1) Multiple cardiac tumours; or 2) A single tumour plus 3) Involvement of other organ systems (central nervous system, kidneys or skin) or a positive family history (5, 7).

The prognosis is rather good, since rhabdomyoma has a tendency to diminish in size during the course of a few years (2, 8). In our case, after delivery there was no sign of heart failure, but arrhythmias were observed. In cases of heart failure, more radical measures should be undertaken (2, 5, 8).

Histological lesions in rhabdomyoma are described as myocardial fibers forming the tumour-like nodules distended with glycogen. Rhabdomyomas may originate in the atria or in the free wall of the ventricular myocardium, but most frequently arise from the ventricular septum. US reveals that most tumours are multiple and solid in character with homogeneous echogenicity, contrary to fibromas which contain multiple areas of calcification and cystic degeneration, which, however, in some cases could not be distinguished from rhabdomyoma (3, 5). Fibromas are usually diagnosed in another, older age group of patients than in our case. It should also be mentioned that teratomas, which can occur in childhood, are mostly extracardiac (5). An additional argument in favour of rhabdomyoma in our case is its increase in size during the foetal period with a tendency to regression during infancy (2, 3, 8).

Antenatal and perinatal US are valuable in cases of intracardiac tumours and are useful in diagnosis of brain abnormalities as quick, non-invasive, cheap techniques prior to CT or MR.

In cases suspected for tuberous sclerosis, spiral CT and/or MR are indicated to confirm lesions which might be present in other organs like kidneys, liver, and bones (1).

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