Case reports

Cardiac rhabdomyomata as a marker for the antenatal detection of tuberous sclerosis

D C CRAWFORD, C GARRETT, M TYNAN, B G NEVILLE, AND L D ALLAN
Departments of Paediatrics and Medical Genetics, Guy's Hospital, London SE1 9RT.

SUMMARY We report the echocardiographic identification of cardiac tumours in antenatal life in a pregnancy where the father was known to have tuberous sclerosis. This allowed termination of an affected pregnancy in the second trimester.

Tuberous sclerosis is an autosomal dominant condition of varying penetrance, in which tuberose deposits are found mainly in the brain but also in the heart, kidneys, and lungs. The associated neurological problems of epilepsy, mental retardation, and occasional motor deficit are caused by the presence of tubers and other defects in cerebral development. The tubers become calcified and are detectable on computerised tomographic scanning of the head. Cardiac manifestations of the disease include multiple rhabdomyomata, which may cause arrhythmias and sudden death, or be large enough to be obstructive.1,2 Frequently cardiac, renal, and lung lesions are asymptomatic and discovered at necropsy. There are also skin manifestations of the disease in the form of shagreen patches or adenoma sebaceum.

We report the echocardiographic identification of cardiac tumours in antenatal life in a pregnancy where the father was known to have tuberous sclerosis. This allowed termination of an affected pregnancy in the second trimester.

Family case report

The proband presented on the first day of life with signs of cyanotic congenital heart disease. Cross-sectional echocardiography demonstrated a tumour completely obstructing the right ventricular cavity and multiple tumourous deposits in the left ventricle. Partial resection of the obstructive tumour was attempted but the child died. Necropsy confirmed the echocardiographic findings of rhabdomyomata and also demonstrated the typical intracranial lesions associated with tuberous sclerosis. The rest of the family was then investigated. The elder sib had epilepsy, severe mental retardation, adenoma sebaceum, and a shagreen patch. Periventricular calcification and dilatation of the left lateral ventricle was evident on computerised tomography of the head. Cross-sectional echocardiography demonstrated the presence of multiple rhabdomyomata of the heart which were small and not obstructive. The father had had epilepsy in childhood and made slow progress at a normal school. He had obvious adenoma sebaceum and CT scanning showed

![Echocardiogram](http://jmg.bmj.com/)

**FIGURE** Echocardiogram of the fetal heart during the 22nd week of pregnancy. A large tumour is seen within the body of the right ventricle and another tumour on the anterior surface of the right atrium.
304

Unusual ocular findings in an infant with cri-du-chat syndrome

SOFIA KITSIOU-TZELI*, H D DELLAGRAMMATICAL*, C B PAPAS*, I D LADAS†, AND C S BARTSOCAS*

*Second Department of Pediatrics, University of Athens, 'P and A Kyriakou' Children's Hospital; and
†Department of Ophthalmology, University of Athens, Greece.

SUMMARY A newborn male with cri-du-chat syndrome, congenital nuclear cataracts, microspherophakia, and probably ectopic lenses is reported. Microspherophakia in cri-du-chat syndrome has not been previously described. The congenital cataracts were inherited from his mother who had a balanced 5;13 translocation; the two events are considered to be coincidental and a possible 'position effect' was excluded, since the other members of her family with congenital cataracts, were chromosomally normal. This is the fourth case reported where familial cri-du-chat syndrome involves chromosomes 5p and 13q.