Tuberous Sclerosis
Cardiac Considerations

Typical cardiac abnormality: rhabdomyoma

Most common cardiac tumor in infants and children

• sl > fibroma in pediatric series
• myxoma is more common in adults
### Tuberous Sclerosis
#### Cardiac Considerations

**Presentation of cardiac tumor**

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<th>Symptom</th>
<th>Diagnosis</th>
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<td>conduction block</td>
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<td>a gallop</td>
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<td>pulm embolus</td>
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<td>SVT</td>
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<td>sudden cardiac death</td>
<td>PS</td>
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**Typical cardiac abnormality: rhabdomyoma**

³/₄ of cases identified < 1 year old
Tuberous Sclerosis
Cardiac Considerations

Typical cardiac abnormality: rhabdomyoma

Histologically benign

May be physiologically malignant

Can occur in fetus or newborn

Tuberous Sclerosis
Cardiac Considerations

Typical cardiac abnormality: rhabdomyoma

- usually multiple
- RV=LV=Septal location frequency
- several mm to several cm
- may be larger than the ventricular cavity
- > ½ of tuberous sclerosis patient have rhabdomyoma
- 80% of cardiac rhabdomyomas have TS
Typical cardiac abnormality: rhabdomyoma

- Almost 50% of tuberous sclerosis patients without signs or symptoms of heart disease will have one or more rhabdomyomas by echocardiography.

The majority of cardiac rhabdomyomas cardiac regress spontaneously, therefore, surgery is not routinely required.

(“responsive to radiation story”)
Cardiac tumors may cause hemodynamic abnormalities

- RV inflow obstruction
- RV outflow obstruction
- LV inflow obstruction
- LV outflow obstruction

*Figure 13:* Multiple rhabdomyomas of the heart. Right atrial, right ventricular view. Arrows point to the large tumor at the junction of anterior and septal leaflets of the tricuspid valve. RA = right atrium; RV = right ventricle.
RVOTO from rhabdomyoma

Bharati and Lev
Futura, 1996

Mitral valve (LV filling) obstruction from rhabdomyoma
Bharati and Lev
Futura, 1996
Cardiac tumors may cause arrhythmias

Ventricular arrhythmia
PVC’s
V tach
Conduction block (e.g., mesothelioma)
Supraventricular arrhythmia
SVT
WPW
AET
Sudden cardiac death

Figure 11: Rhabdomyoma of the left ventricle obstructing the outflow tract into the aorta. LA = left atrium; LV = left ventricle. Arrows point to the rhabdomyoma obstructing the outflow tract of the left ventricle.
Cardiac tumors may cause arrhythmias

Ventricular arrhythmia
PVC’s
V tach

Purkinje cell tumor

Subset of rhabdomyoma
Intractable arrhythmia
“MAP and DESTROY”
Total A-V block due to tuberous sclerosis. A case report
Valente N, Guidugli Neto J, De Paulo AA, Pimenta J

Tuberous sclerosis is a neurologic disease affecting various organs with a triade: sebaceum adenoma, mental retardation and seizures. This report presents a case of a patient with tuberous sclerosis and third degree A-V block with complete invasive and non-invasive evaluation. The patient had sincope and complete A-V block with QRS complexes showing right bundle branch block morphology. The echocardiogram showed dilated cardiomyopathy with diffuse left ventricular dysfunction and had normal coronary arteriography. The electrophysiologic evaluation showed complete infra-hisian A-V block and QRS with left bundle branch block pattern with normal sinus nodal and A-V nodal function. It was not possible to induce ventricular tachyarrhythmias up to two extrastimuli. Histologic study showed normal myocardium under light and electronic microscopy. After permanent VVI pacemaker implant, the patient in follow-up for 16 years. This case seems to be the first in the international medical literature of tuberous sclerosis with complete heart block.

Rhabdomyoma and ventricular preexcitation syndrome. A report of two cases and review of literature.
Mehta AV

Department of Pediatrics, James H. Quillen College of Medicine, East Tennessee State

Two newborn female infants with ventricular preexcitation syndrome, supraventricular tachycardia, rhabdomyomatous tumor of the heart, and tuberous sclerosis are described. The first patient had unsuccessful partial resection of the rhabdomyomatous tumors obstructing the tricuspid valve and right ventricular cavity and died immediately after surgery. By histologic examination, no direct accessory connection was noted between the myocardial fibers of atria and ventricles through annulus fibrosus. By gross examination, the tumor extended from the right atrium through the tricuspid valve to the right ventricular cavity, suggestive of macroscopic accessory connection. The second patient presented with unsustained ventricular tachycardia and obstructive subaortic rhabdomyoma, requiring emergency surgery. One week later, reentrant supraventricular tachycardia developed and she required digoxin therapy for 15 months. CONCLUSIONS: Infants with rhabdomyomatous tumor of the heart and ventricular preexcitation syndrome may have microscopic or macroscopic accessory connections. Cardiac tumors like rhabdomyoma and oncocyctic tumors should be suspected in infants with ventricular preexcitation syndrome or supraventricular tachycardia.
Subset of adult women with severe and rapidly progressing pulmonary disease

Gomez, MR

*Tuberous Sclerosis*
New York, Raven Press 1979

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De Rosa N

Pulmonary lymphangioleiomyomatosis (PLAM) is a rare disease that exclusively affects *young women of reproductive age*. It is characterized by widespread pulmonary proliferation of abnormal, *immature smooth muscle cells (lam cells) leading to cystic destruction of the lung parenchyma*. Lam occurs frequently in the thoracic duct and in axial lymph nodes, mediastinal or retroperitoneal. It can occur either *in association with tuberous sclerosis complex or without* TSC. A case of TSC-LAM is reported, and the histogenesis and the role of lymphangiogenesis in the progression of disease is discussed.
Angiomyolipoma of kidney and cystic renal disease

Late hypertension

Arterial aneurysms and tuberous sclerosis: a classic but little known association.
Calcagni G, Gesualdo F, Tamisier D, Brunelle F, Sidi D, Ou P

Department of Pediatric Cardiology, Hôpital Necker Enfants Malades, AP-HP, University Paris Descartes, Paris, France.

We report a 20-month-old boy with tuberous sclerosis and iliofemoral arterial aneurysms. This case highlights the importance of systematic screening for such vascular complications. Multislice CT or MRI is mandatory as a screening and diagnostic tool in this setting to complement US with Doppler examination.
Cerebral and renal embolic lesions have occurred from cardiac rhabdomyomas in tuberous sclerosis

Kandt et al
*Neurology*, 1985 35:1223-1225