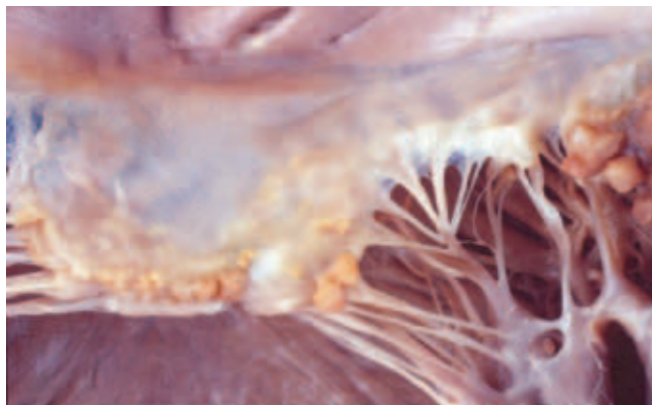


### Echocardiographic Findings

As mentioned above, pericardial involvement is most common; therefore, pericardial effusions +/- pericardial thickening are often seen. Valvular thickening and valvular regurgitation are also commonly seen. When present, Libman-Sacks vegetations appear as heterogeneous, echogenic lesions that are round with irregular borders and usually less than 1 cm<sup>2</sup> in size (Fig. 14.5); lesions are attached to the atrial surface of the mitral valve and the aortic or ventricular side of the aortic valve and display no independent motion.

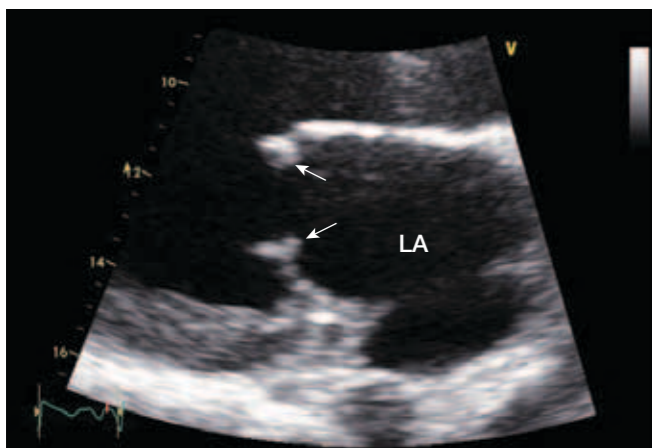
Libman-Sacks vegetations can be found in approximately 1 in 10 patients (or 10% of patients) with SLE. The most frequently involved valve is the mitral valve followed by the aortic valve. Patients who have Libman-Sacks vegetations have a longer disease duration and higher disease activity. Furthermore, these patients are at greater risk of cerebral ischaemic events than those without these lesions.

**Source:** Moyssakis I, Tektonidou MG, Vasiliou VA, Samarkos M, Votteas V, Moutsopoulos HM. Libman-Sacks endocarditis in systemic lupus erythematosus: prevalence, associations, and evolution. *Am J Med.* 2007 Jul;120(7):636-42.



**Figure 14.4** This gross pathological specimen from a patient with systemic lupus erythematosus shows Libman-Sacks endocarditis of the mitral valve. Observe the characteristic multiple wart-like, tan masses on the atrial side of the valve; these lesions may occur along the free edge or closing edge of the mitral valve, the ventricular aspect of the leaflets, or on the tendinous cords.

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**Figure 14.5** Libman-Sacks vegetations of the mitral valve are shown on a zoomed parasternal long axis view. Observe the nodular, echogenic masses on the left atrial (LA) side of the anterior and posterior mitral leaflets (arrows).

Other cardiac abnormalities that may be detected on the echocardiographic examination include:

- signs of pulmonary hypertension (for example, increased tricuspid regurgitant velocity),
- RWMA secondary to myocardial infarction.

#### Libman-Sacks versus Infective Endocarditis

Patients with SLE may have some clinical features consistent with infective endocarditis (IE); therefore, Libman-Sacks endocarditis may be confused with IE. Libman-Sacks endocarditis can be differentiated from IE based on the echocardiographic features of the lesion as cited below. It is important to note, however, that IE can coexist with Libman-Sacks vegetations.

	Infective Endocarditis	Libman-Sacks Endocarditis
Location	Almost always located at the line of leaflet closure	Predominantly located near the leaflet base
Appearance	Usually homogeneous soft tissue density	Usually heterogeneous echodensity with central regions of high reflectance suggesting connective tissue or even calcific density
Mobility	Vibratory or rotatory motion at least partly independent of the motion of the valve	Do not display independent motion; masses exactly parallel motion of related leaflet

**Source:** Roldan CA, Shively BK, Lau CC, Gurule FT, Smith EA, Crawford MH. Systemic lupus erythematosus valve disease by transesophageal echocardiography and the role of antiphospholipid antibodies. *J Am Coll Cardiol.* 1992 Nov 1;20(5):1127-34.

### Hereditary Connective Tissue Disorders

As the name suggests, connective tissue disorders are diseases featuring abnormalities involving the connective tissues, most commonly collagen and elastin. There are more than 200 disorders that affect connective tissues including systemic rheumatic diseases as discussed above. This section focuses on selected hereditary connective tissue disorders.

Hereditary connective tissue disorders are diagnosed based on physical findings and the identification of the causative genetic mutation (Table 14.2). In particular, these syndromes are autosomal-dominant diseases; this means that the offspring of an affected parent have a 50% risk of inheriting the disease (Fig. 14.6). Therefore, echocardiographic examinations are frequently requested for all first-degree relatives of patients with hereditary connective tissue syndromes.

The most common cardiovascular manifestation of these disorders is dilatation of the aorta.

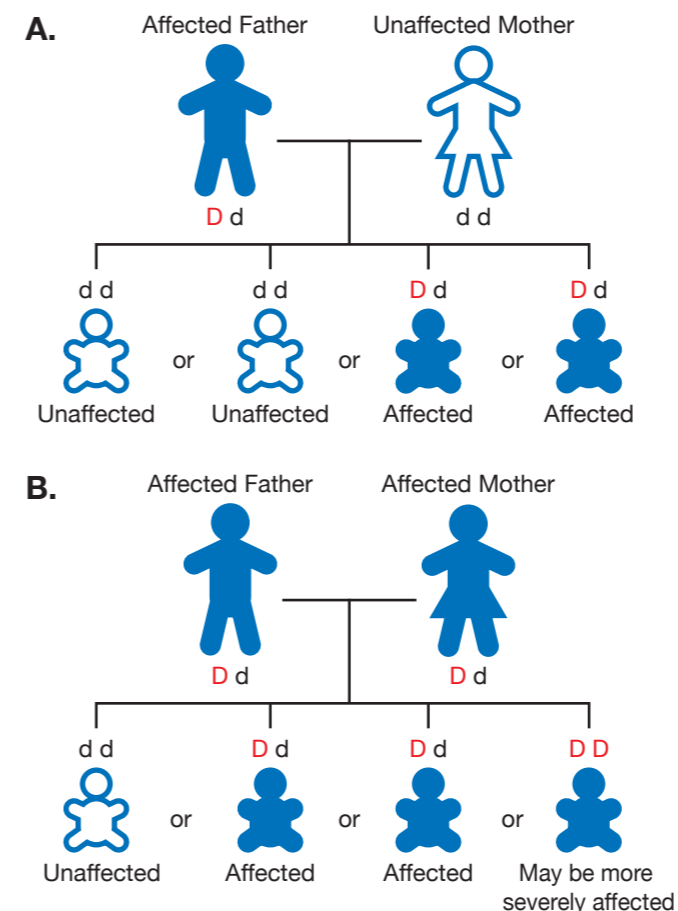
### Marfan Syndrome

#### Overview

Marfan syndrome (MFS) is the most common of the hereditary connective tissue disorders. This syndrome is characterised by skeletal, cardiovascular and ocular features (Table 14.2). Approximately 75% of cases are familial (inherited as an autosomal dominant trait) while 25% of cases are sporadic (that is, the mutation occurs spontaneously and may be associated with older paternal age).

The diagnosis of this syndrome is based on the family history, clinical findings and identification of the defective gene. Clinical findings are based on the Ghent criteria which identify major and minor manifestations in a number of organ systems including the skeletal, ocular, cardiovascular, pulmonary, dura, and integumentary systems<sup>[14.1]</sup>.

Cardiac manifestations of MFS are listed in Table 14.1. In particular, a loss of elastic tissue in the aortic wall results in medial degeneration which weakens the aortic walls leading to aortic dilatation and aortic aneurysms (mostly at the level of the sinuses of Valsalva), and possible type A aortic dissections.



**Figure 14.6** These schematics illustrate the mode of inheritance of autosomal dominant disorders. **Panel A** shows how the disorder may be passed on when one parent has the autosomal dominant faulty gene. In this situation, there is a 50% chance that the child will inherit the faulty gene and will therefore be affected by, or be predisposed to developing the disease. **Panel B** shows how the disease may be passed on when both parents have the autosomal dominant faulty gene. In this situation, there is a 75% chance that the child will inherit the faulty gene from one parent and will therefore be affected by, or be predisposed to developing the disease. There is also a 25% chance that the child will inherit the faulty gene from both parents. In this case, the child may be more severely affected by the disease. **D** = faulty dominant gene, **d** = unaffected or normal gene.

[14.1] Loeys BL, Dietz HC, Braverman AC, et al. The revised Ghent nosology for the Marfan syndrome. *J Med Genet.* 2010 Jul;47(7):476-85.

### Echocardiographic Findings

Echocardiography has a very important role in the assessment of MFS. In particular, characteristic echocardiographic features of a dilated aorta in conjunction with mitral valve prolapse (MVP) are often associated with this syndrome (Fig. 14.7). Most importantly, careful and accurate measurements of the aortic dimensions are required. Annual echocardiographic examinations are recommended for all patients with MFS to monitor the size of the aortic root and the annual rate of growth of the aortic dimensions. If the maximal aortic diameter is 4.5 cm or greater, or if the aortic diameter shows significant growth from baseline, more frequent review is required.

Echocardiography is also valuable in the:

- assessment of the degree of MR (secondary to MVP),
- assessment of the aetiology and degree of AR (secondary to aortic root dilatation),
- identification of dilatation of the main pulmonary artery (in the absence of pulmonary valve stenosis),
- identification of mitral annular calcification (< 40 yrs of age),
- diagnosis of a type A aortic dissection.

### Ehlers-Danlos Syndrome

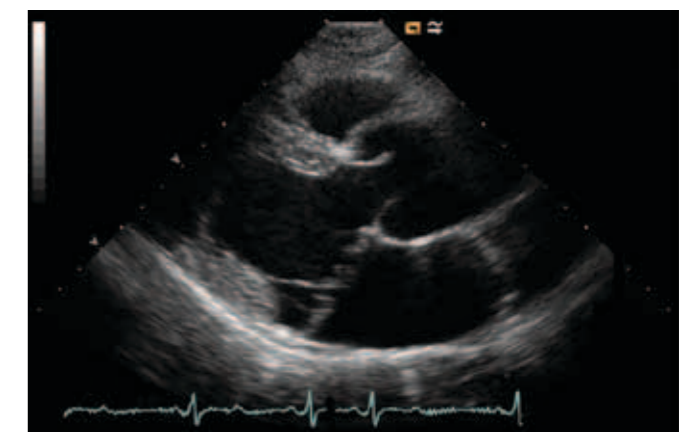
#### Overview

Ehlers-Danlos syndrome (EDS) refers to a group of inherited, connective tissue disorders that are characterised by joint hypermobility, cutaneous fragility and hyperextensibility.

There are more than 10 different types of disorders associated with EDS. The type that is most relevant with respect to cardiovascular involvement is EDS type IV, the vascular type of EDS. This type of EDS is defined by four features: (1) facial acrogeria (emaciated face with prominent cheekbones and sunken cheeks), (2) translucent skin with highly visible subcutaneous vessels on the trunk and lower back, (3) easy bruising, and (4) severe arterial, digestive and uterine complications.

The diagnosis of this syndrome is based on the family history, clinical features and identification of the defective gene.

The pathophysiology and, therefore, the cardiovascular manifestations of EDS type IV are very similar to MFS; these manifestations are listed in Table 14.1.



**Figure 14.7** This image was recorded from a parasternal long axis view in a patient with Marfan syndrome. Observe that there is effacement of the aorta with aortic root dilatation (the aortic root measured 4.5 cm). Bileaflet mitral valve prolapse is also noted on this systolic frame.