

# 15.7

## CLINICAL APPLICATION

### MOLECULAR CAUSES OF CARDIOVASCULAR DISEASE

A variety of inherited and environmental factors contribute to causing cardiovascular disease. Many cases are probably due to a diet high in refined carbohydrates and/or saturated fats and sedentary lifestyle, against a backdrop of genetic predisposition. Disorders of the heart and blood vessels caused by single genes are very rare, but understanding how they arise can provide insights that are useful in developing treatments for more prevalent forms of disease. For example, widely used cholesterol-lowering drugs called statins were developed based on understanding familial hypercholesterolemia, an inherited condition that affects one-in-a-million children.

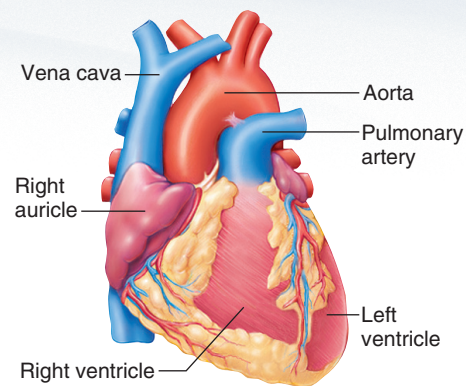
#### A Connective Tissue Defect

In January 1986, volleyball champion Flo Hyman left the court during a game in Japan, collapsed, and died. Her aorta had burst. Hyman had *Marfan syndrome*, an inherited condition that also caused the characteristics that led her to excel in her sport—her great height and long fingers (fig. 15J).

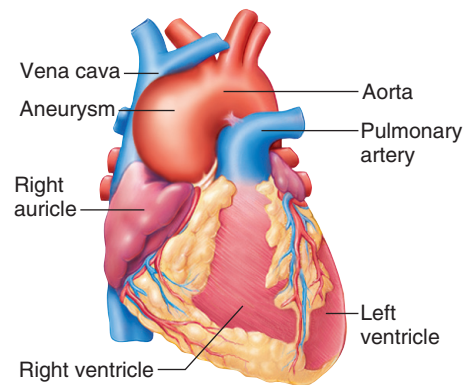
In Marfan syndrome, an abnormal form of a connective tissue protein called fibrillin weakens the aorta wall. After Flo Hyman died, her siblings were examined, and her brother Michael was found to have a weakened aorta. By surgically repairing his aorta and giving him drugs to control his blood pressure and heart rate, physicians enabled him to avoid the sudden death that claimed his famous sister. Testing for the



(a)



Normal heart



Heart in Marfan syndrome

(b)

#### FIGURE 15J

Aneurysm. (a) Two years after she led the 1984 U.S. women's volleyball team to a silver medal in the Olympics, Flo Hyman died suddenly when her aorta burst, a symptom of Marfan syndrome. (b) Note the swelling (aneurysm) of the aorta in the heart of a person with Marfan syndrome. A burst aneurysm here is fatal.