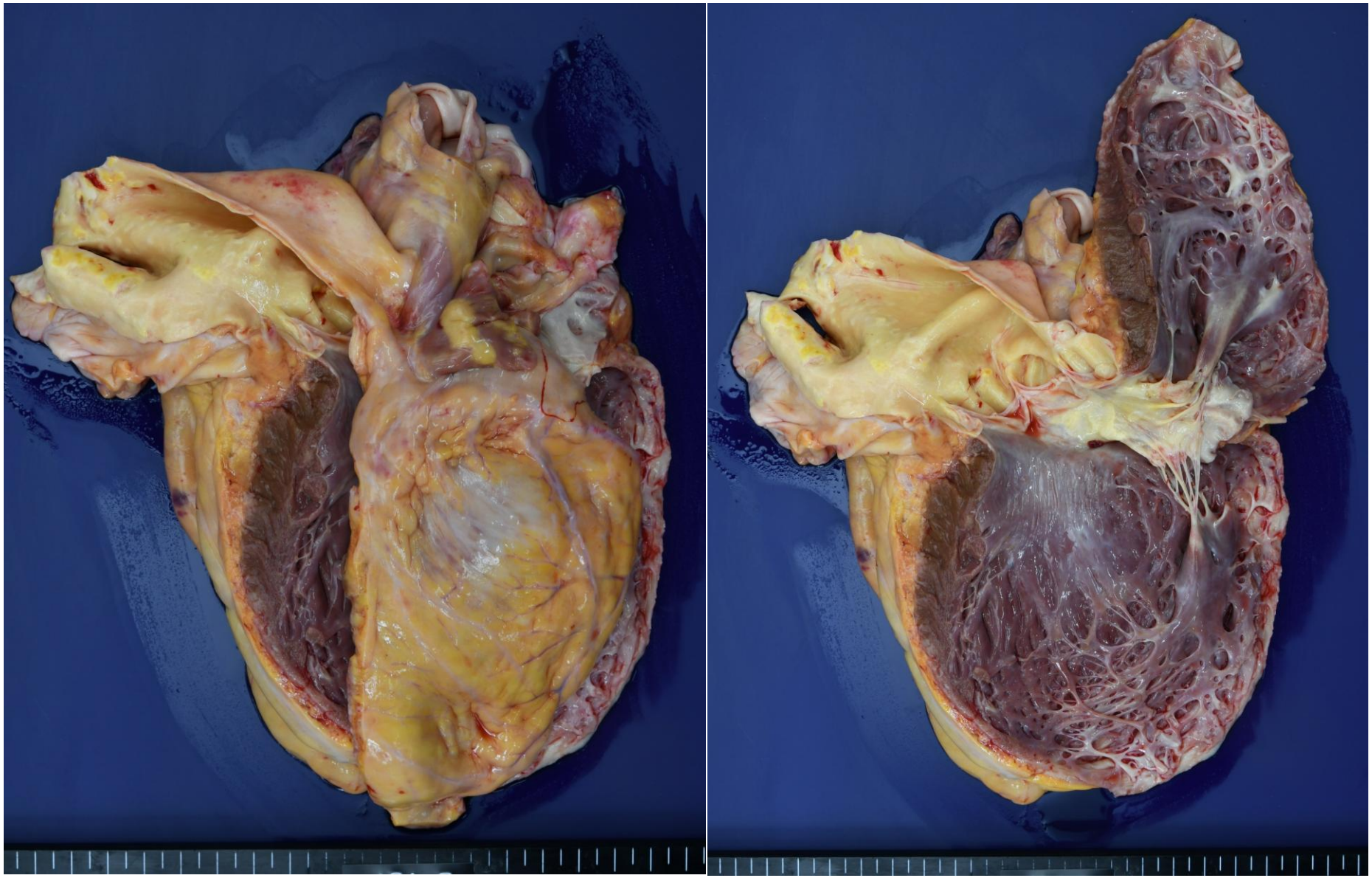
Familial dilated cardiomyopathy

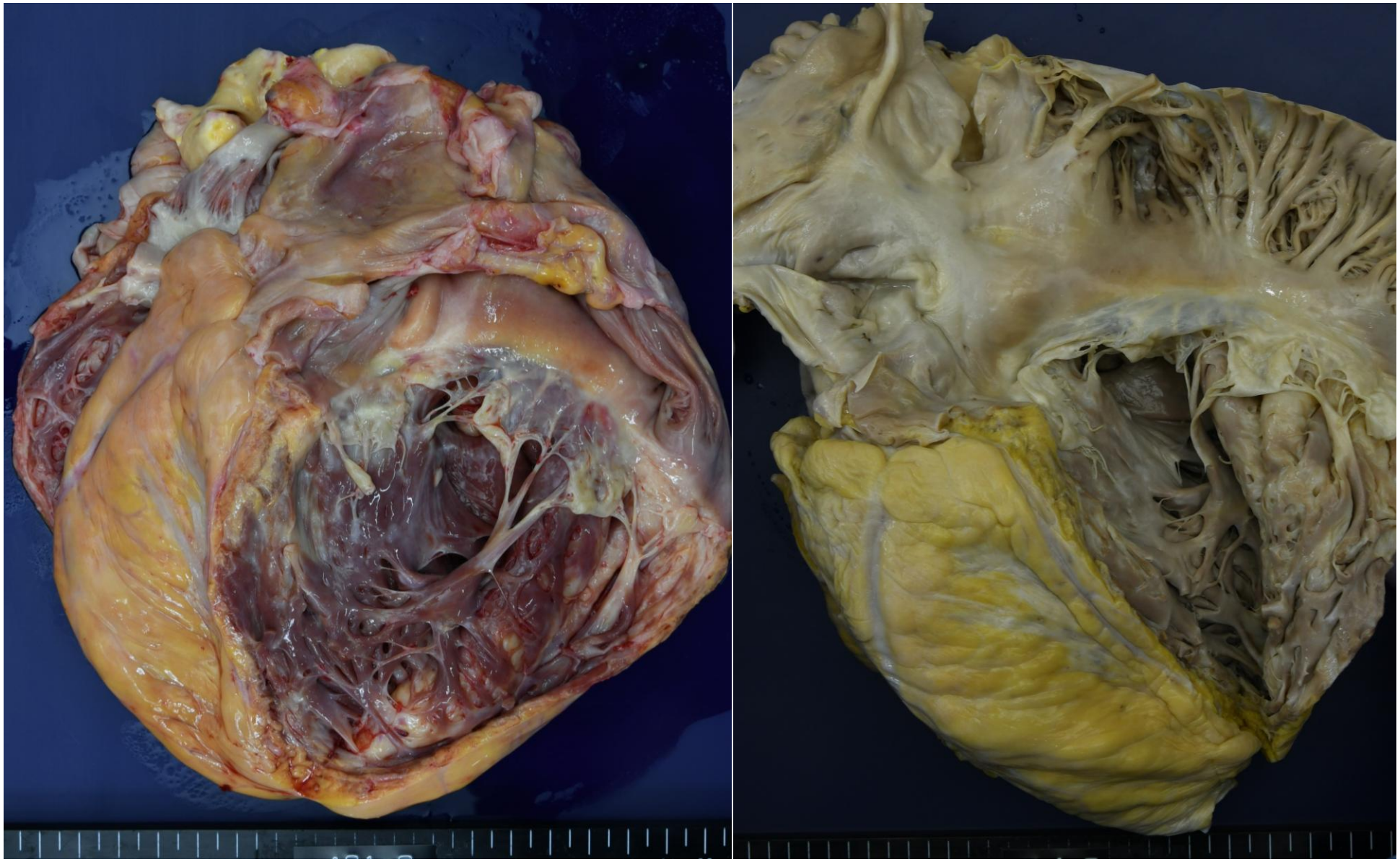
Familial dilated cardiomyopathy is a genetic form of myocardial disease. One-third to one-half of patients with dilated cardiomyopathy have a family history of the disease. Most of the familial cases show autosomal dominant inheritance. More than 100 genes have been implicated to date. The heart shows thinning and weakening of the cardiac muscles, causing marked dilatation of the chamber. The heart becomes unable to pump blood as efficiently as usual, and over time, heart failure is inevitable. The disease typically begins in mid-adulthood, but can occur at any time from infancy to late adulthood. Signs and symptoms of familial dilated cardiomyopathy can include arrhythmia, dyspnea, fatigue, syncope and peripheral edema. The first sign of the disorder may happen as sudden cardiac death. It usually takes many years for symptoms caused by cardiac dysfunctions.

Ref.-1: Peters S, et al. Familial dilated cardiomyopathy. Heart Lung Circ 2020; 29(4): 566-574. doi: 10.1016/j.hlc.2019.11.018

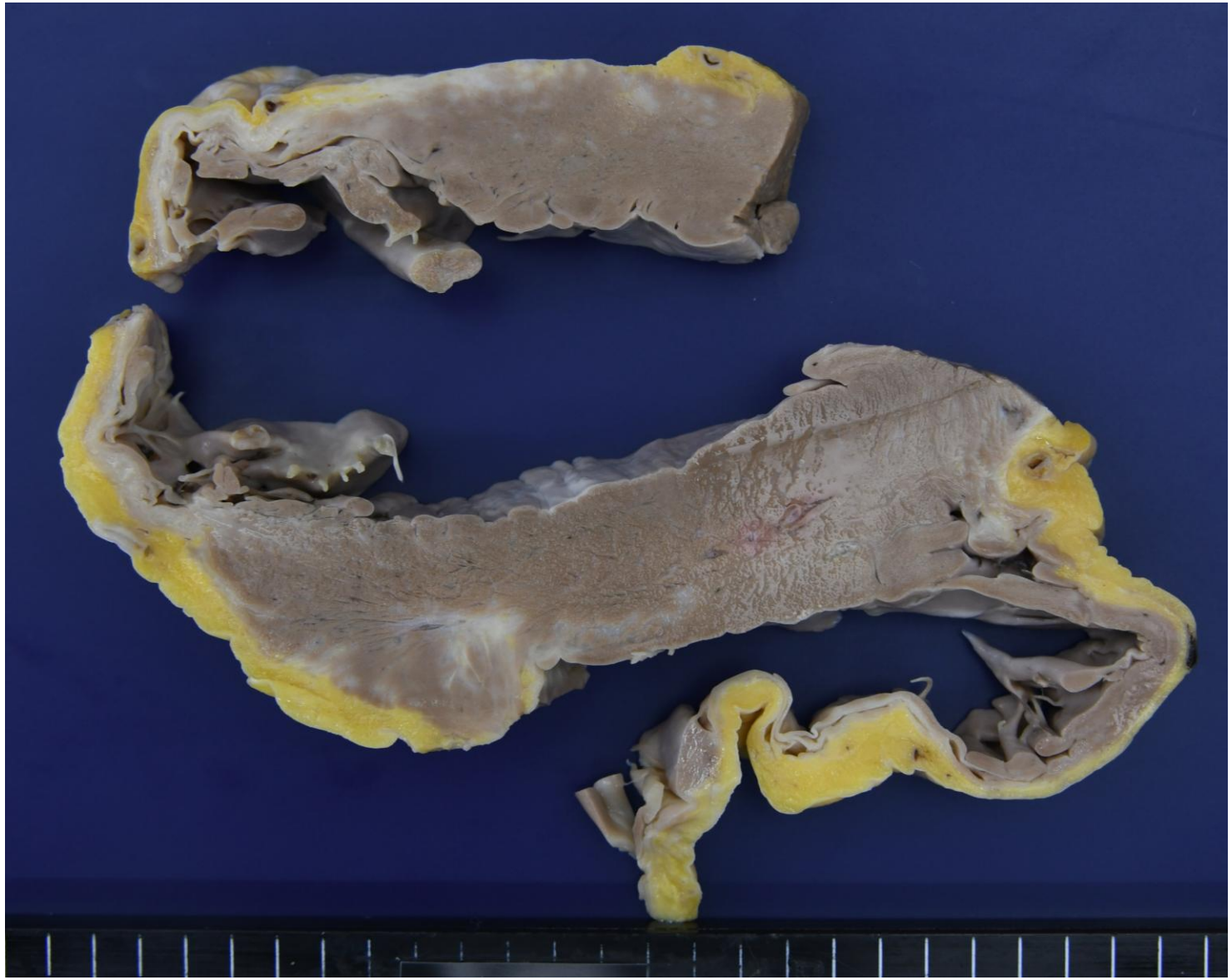
Ref.-2: Mitrut R, et al. Histopathological aspects of the myocardium in dilated cardiomyopathy. Curr Health Sci J 2018; 44(3): 243-249. doi: 10.12865/CHSJ.44.03.07



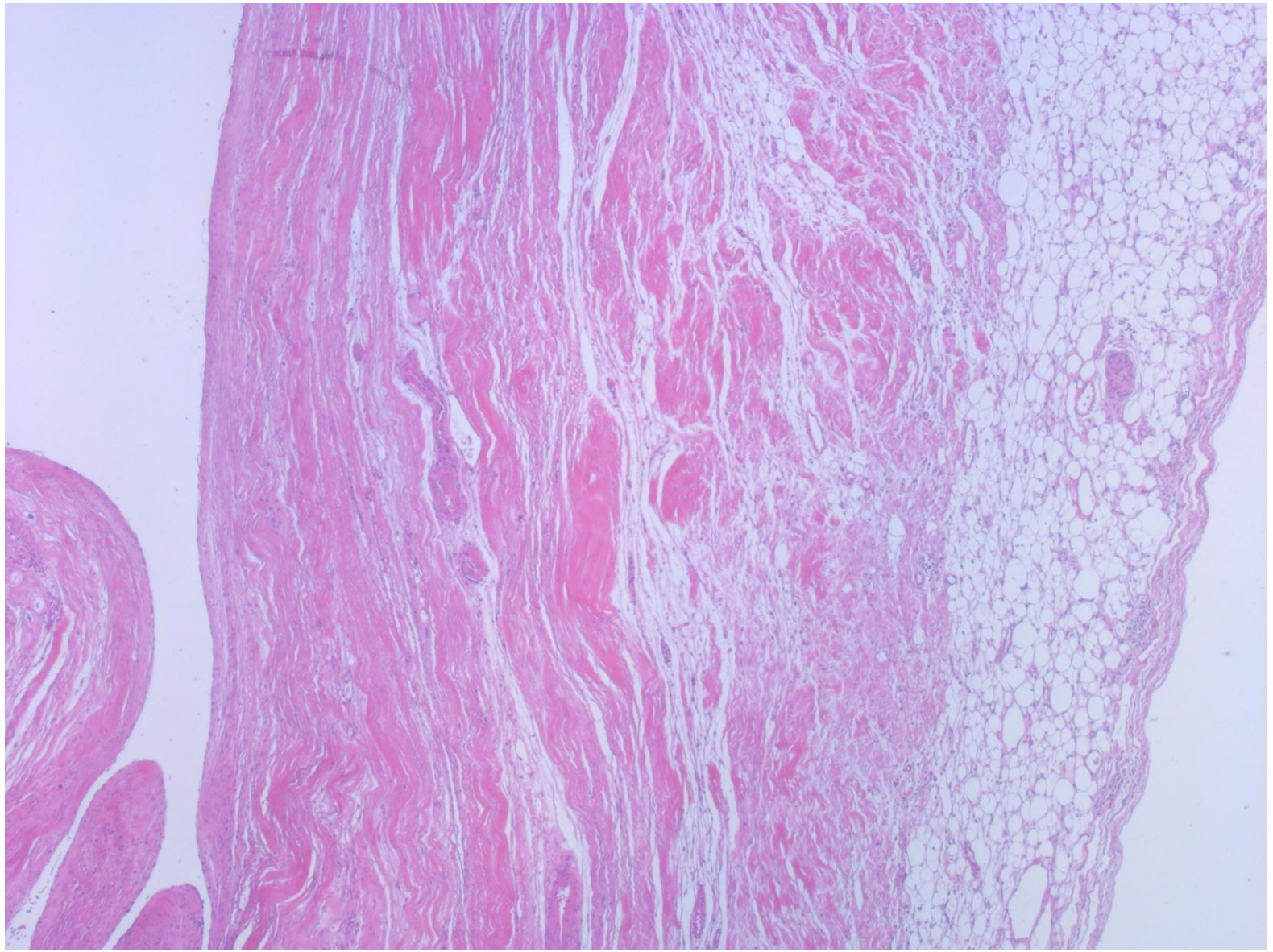
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The heart weighs 480 g. The left ventricle is prominently dilated and the lateral wall is markedly thinned and fibrosed.



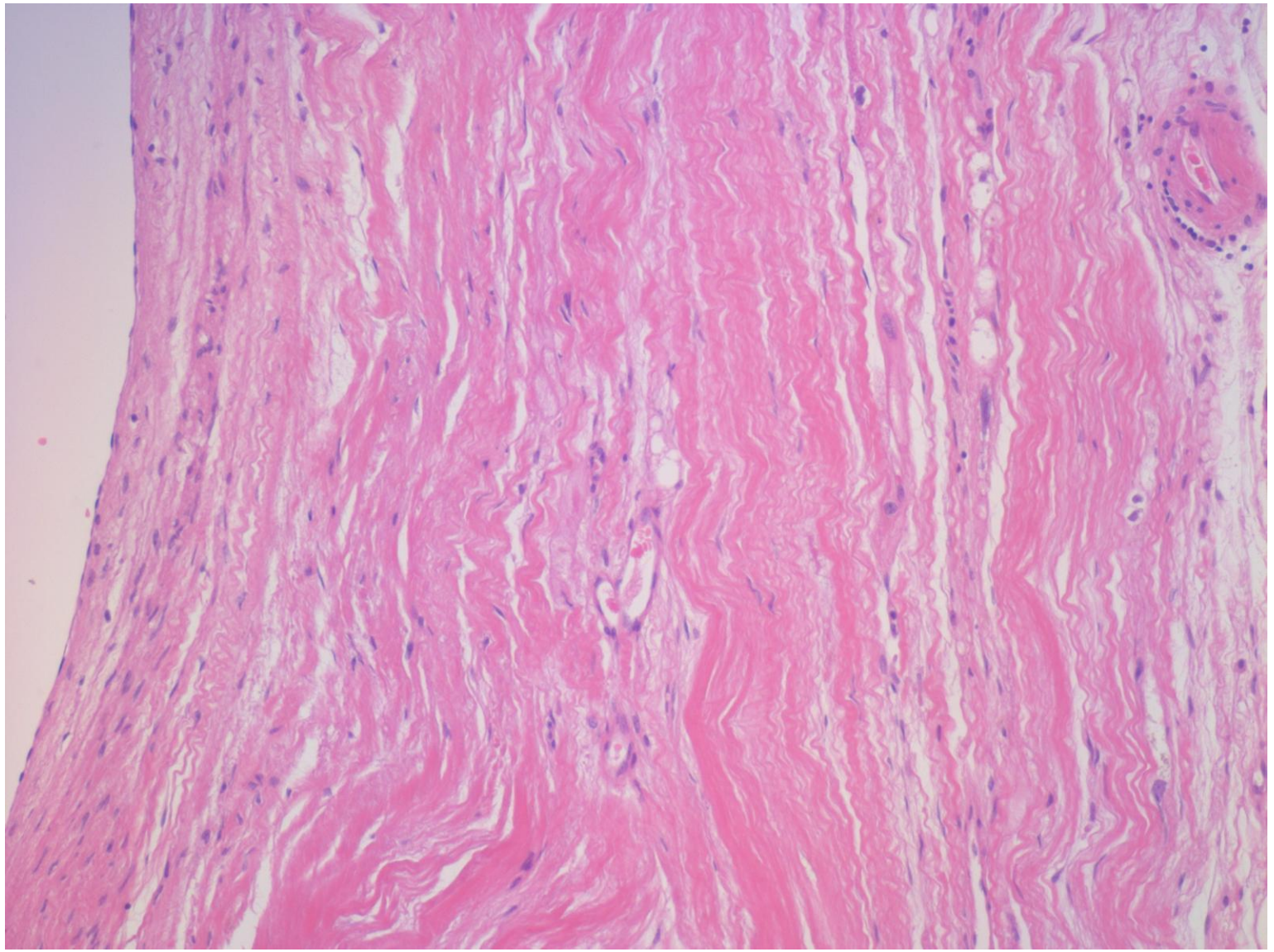
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The heart weighs 480 g. The dilatation of the right ventricle is shown.



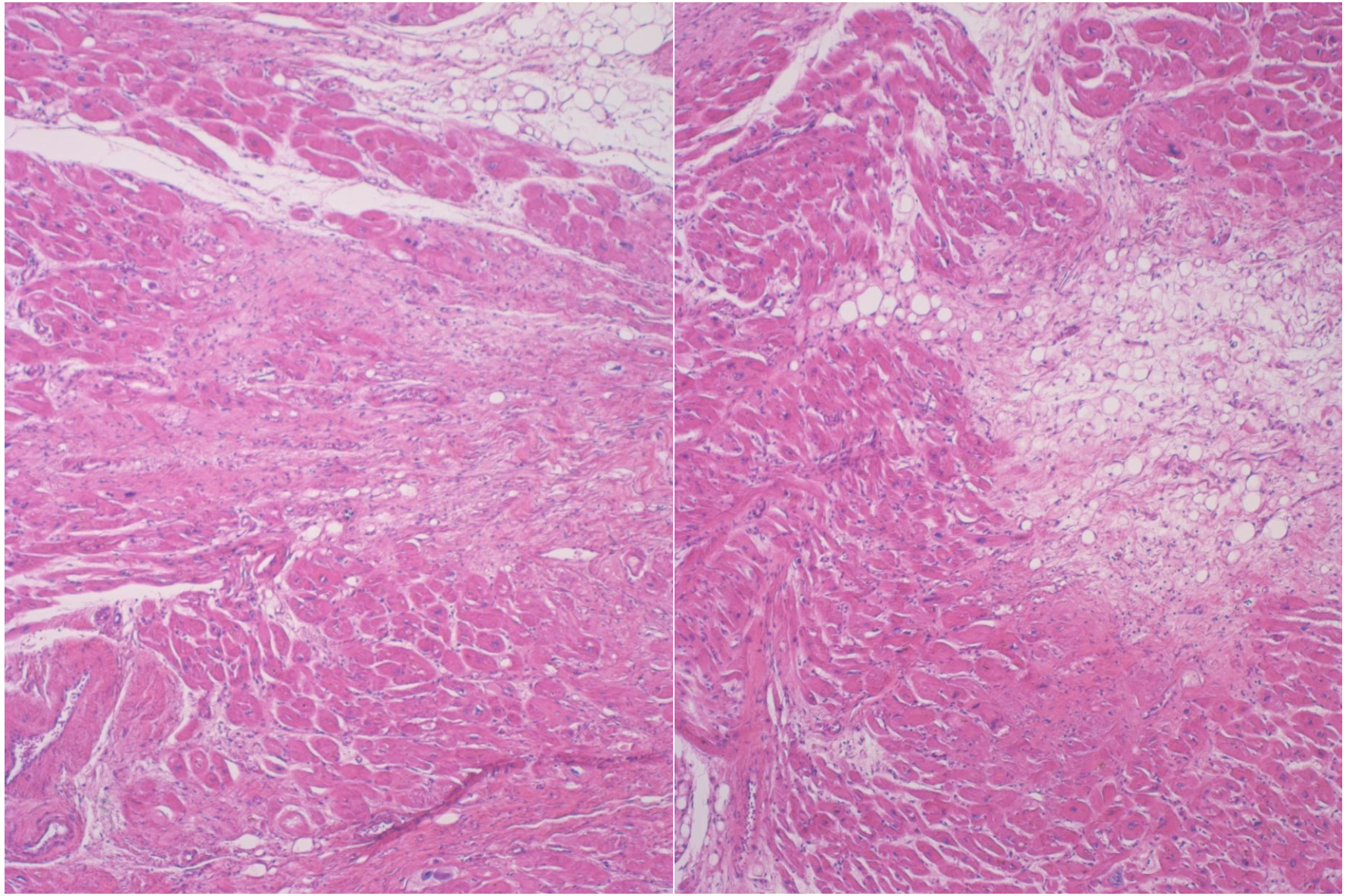
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The horizontal section of the heart shows marked thinning and fibrosis of both the left and right ventricles. The lumina of both ventricles are dilated.



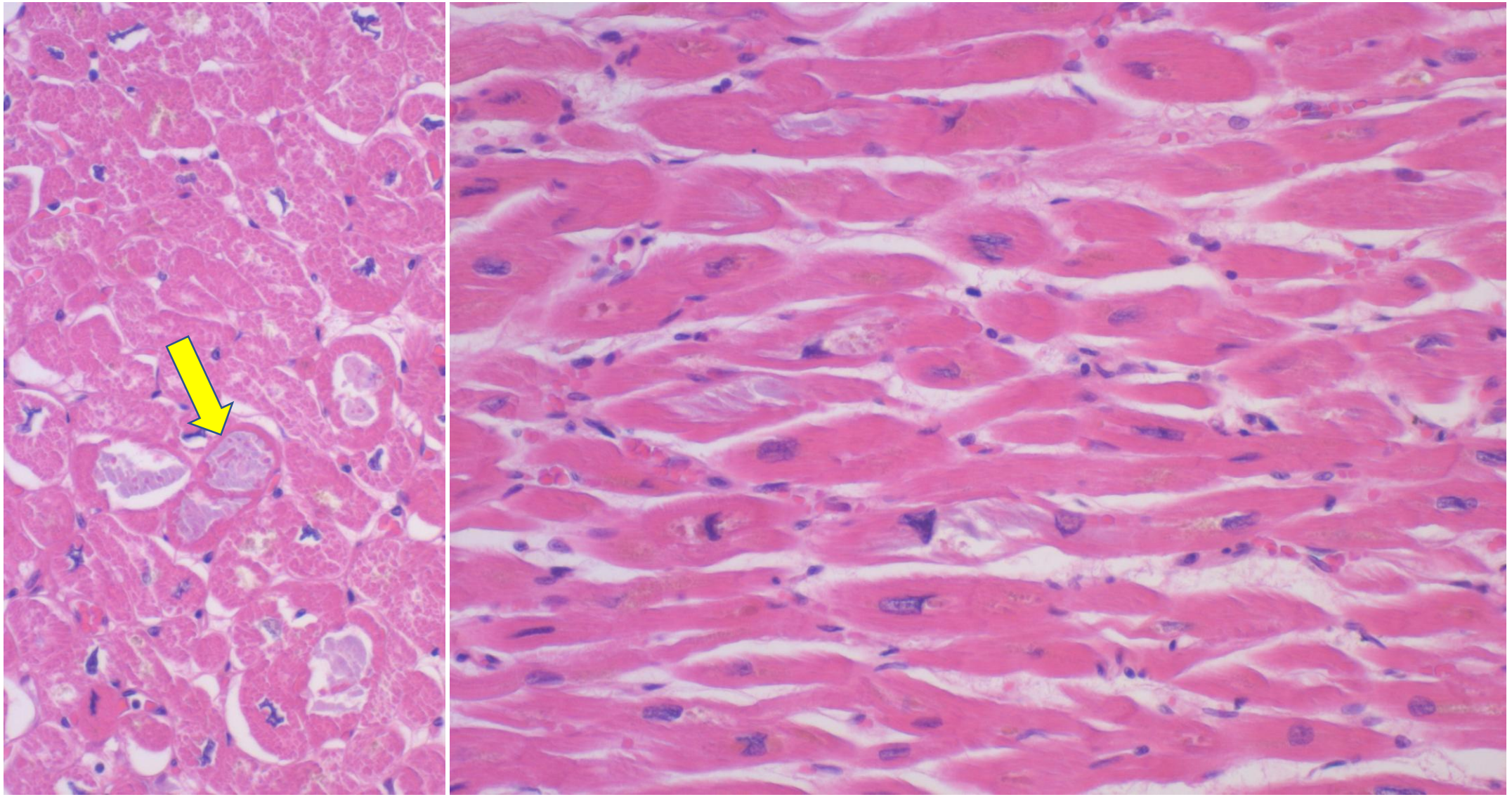
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The lateral wall of the left ventricle shows disappearance of the subserosal cardiac muscle layer replaced by fibrosis (H&E-1).



Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The lateral wall of the left ventricle shows disappearance of the subserosal cardiac muscle layer replaced by dense fibrosis (H&E-2).

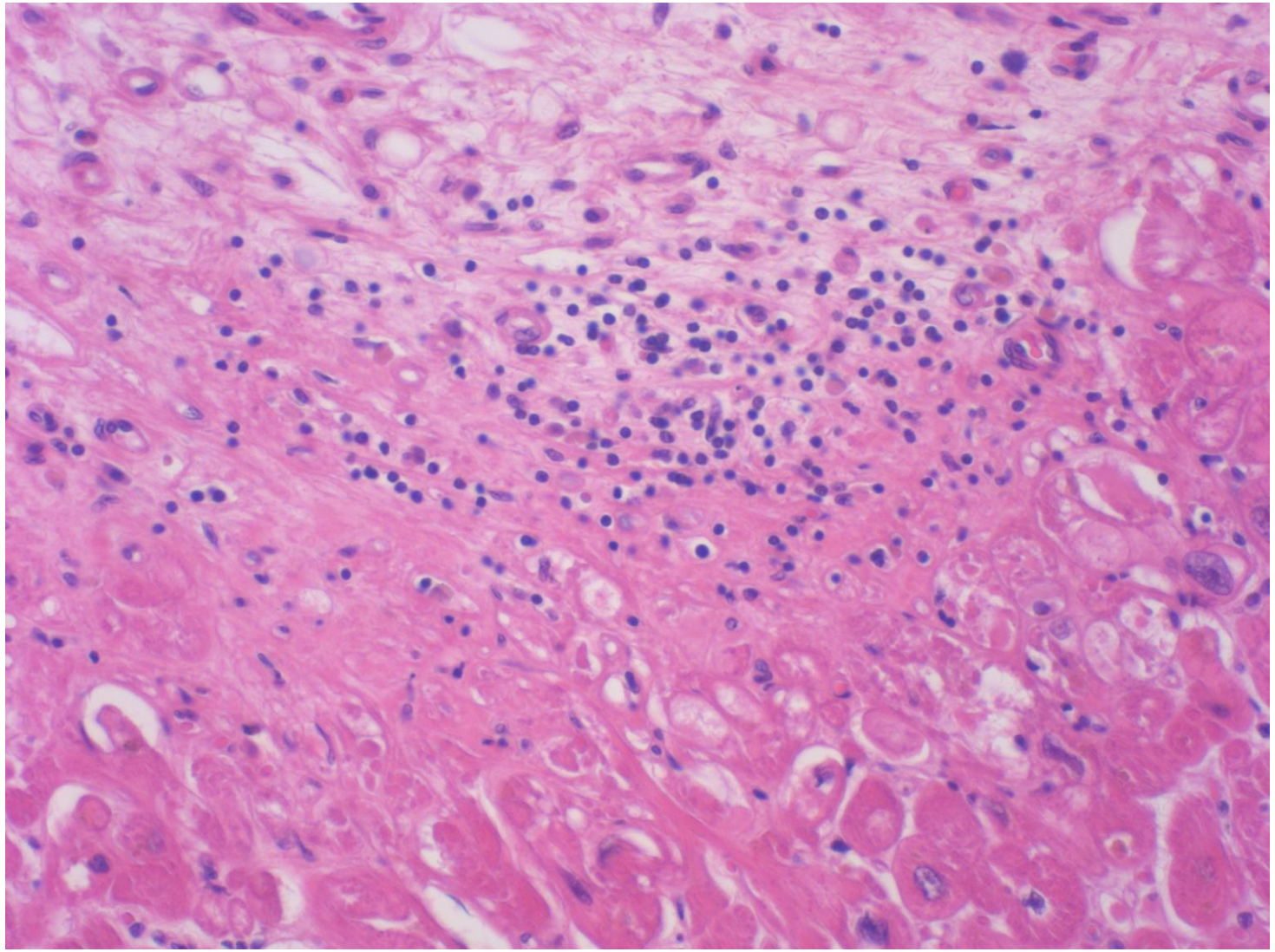


Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The posterior wall of the left ventricle shows patchy loss of the cardiomyocytes with replacement by fibrosis (H&E-3).



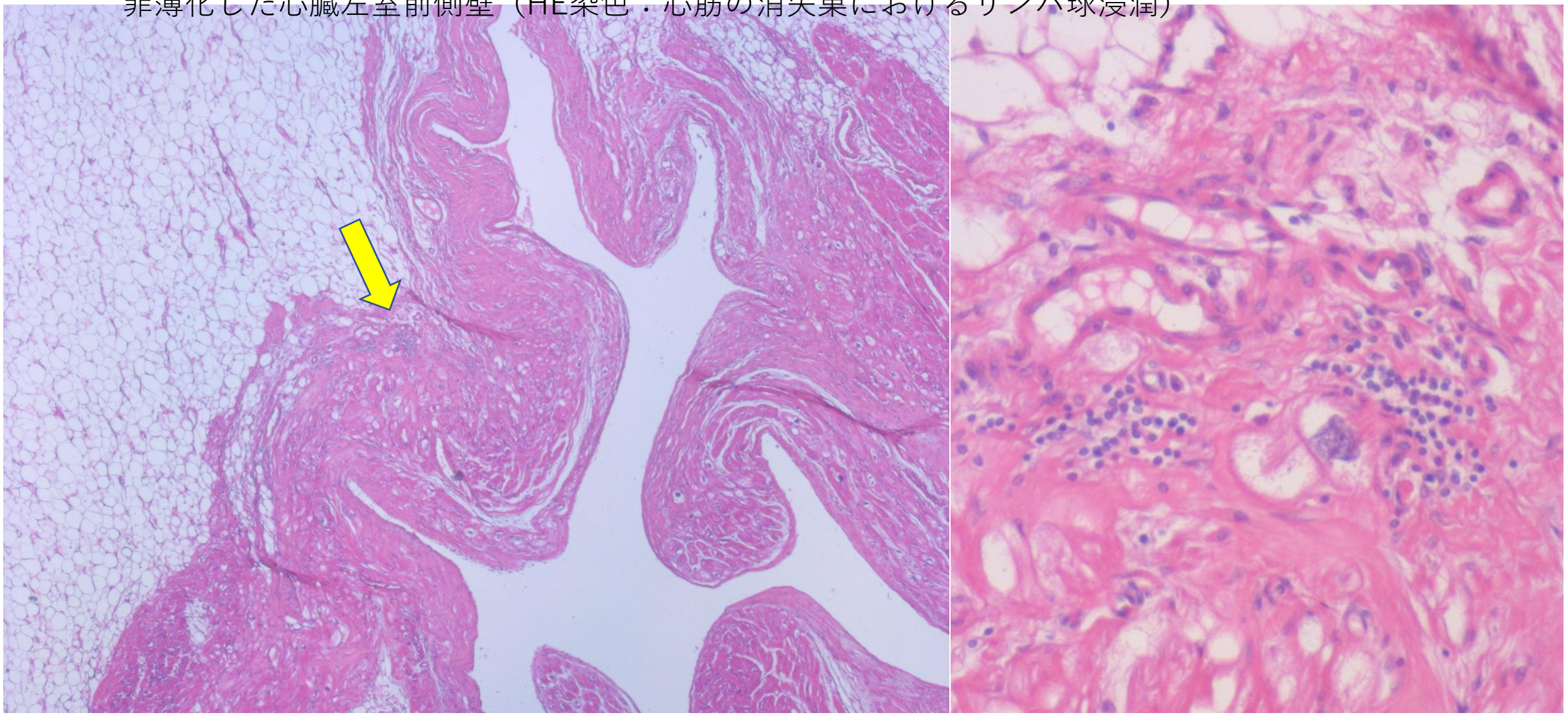
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The anterior wall of the left ventricle shows intracellular deposition of basophilic materials (left: arrow) and hypertrophic cardiomyocytes with box-shaped enlarged nuclei (right) (H&E-4).

心臓左室後壁（HE染色：漿膜側心筋の消失巣におけるリンパ球浸潤）

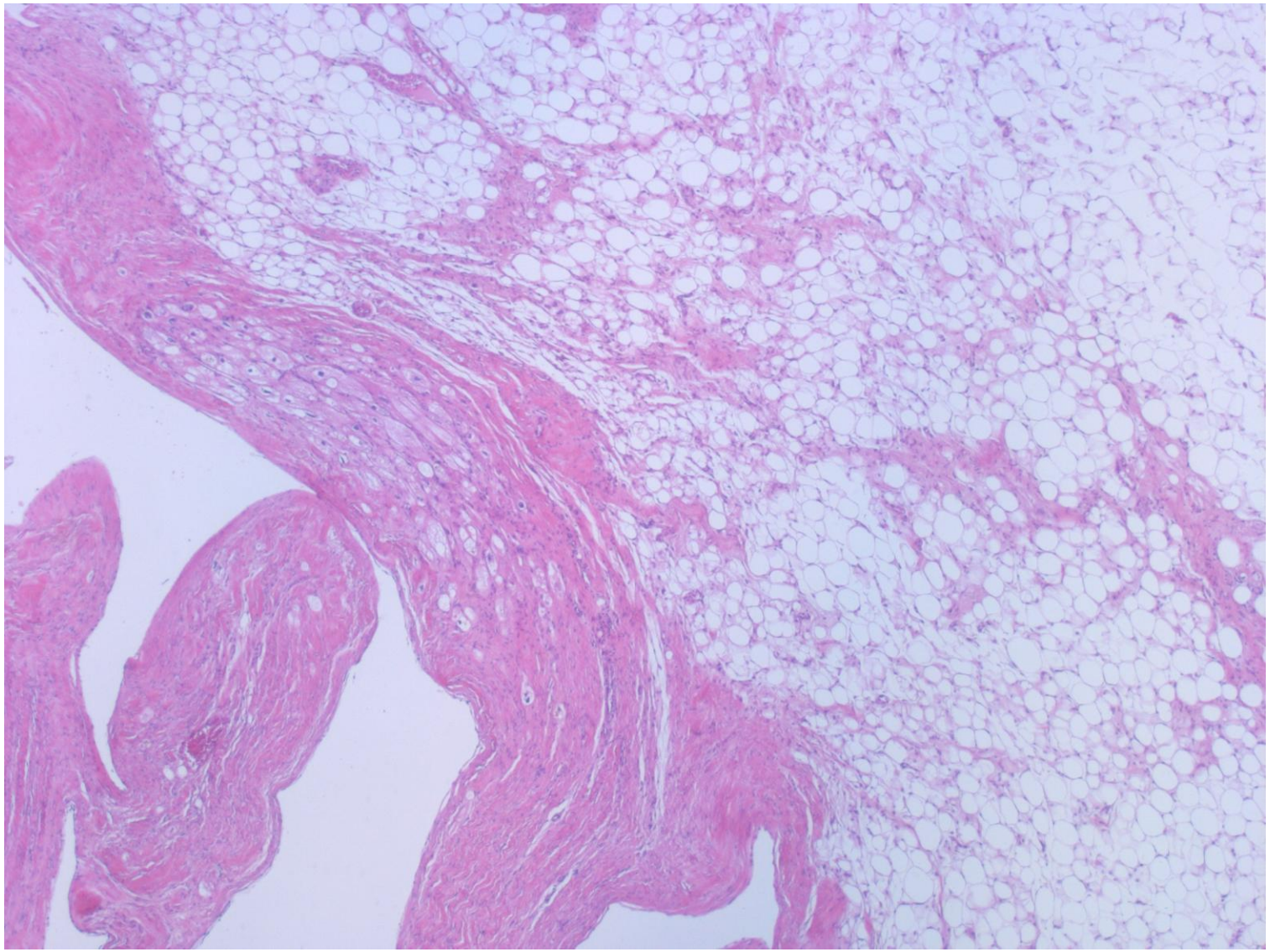


Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The posterior wall of the left ventricle shows focal lymphocytic infiltration around the fibrotic lesion in the subserosal area (H&E-5).

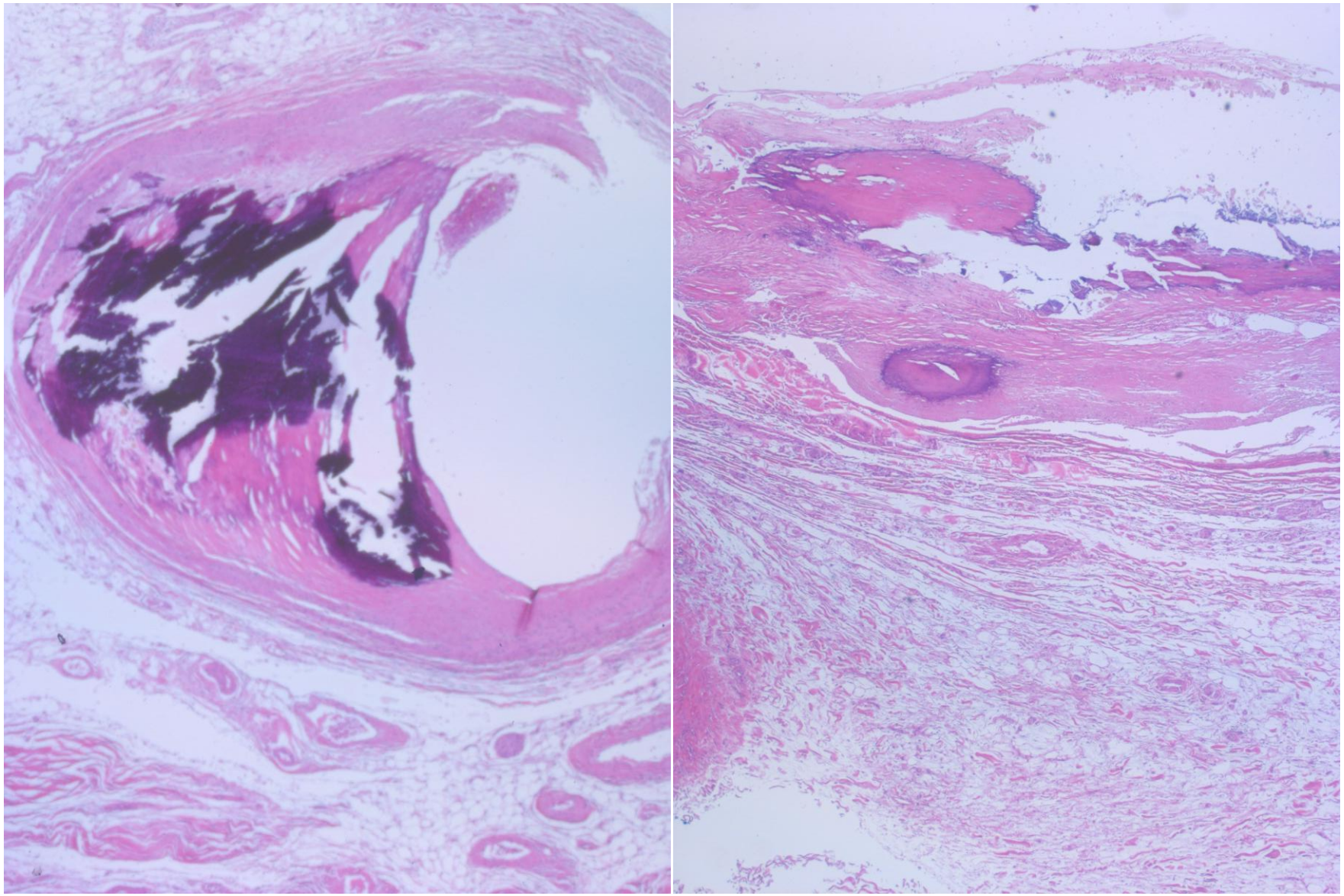
菲薄化した心臓左室前側壁（HE染色：心筋の消失巣におけるリンパ球浸潤）



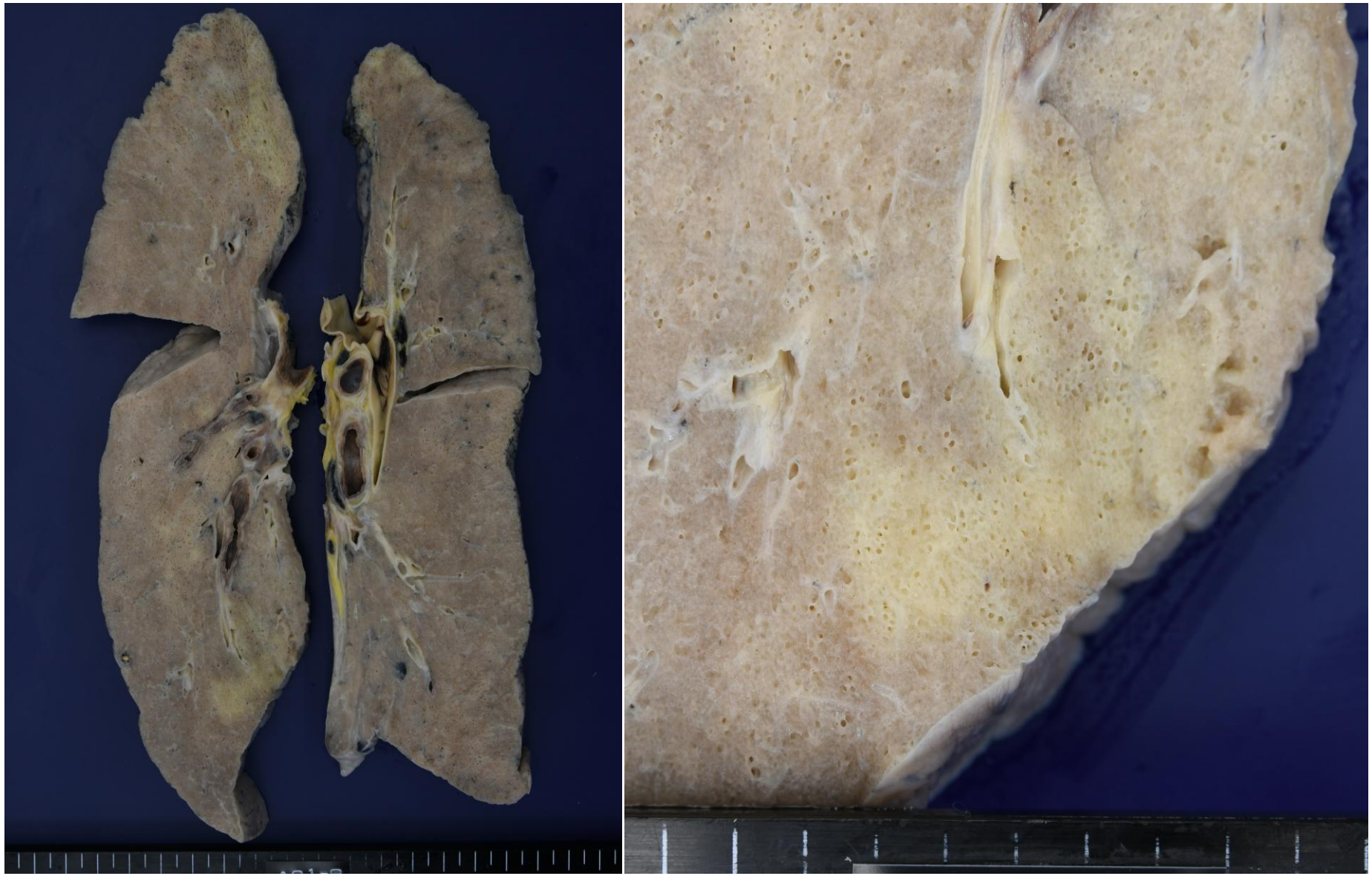
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The thinned anterior wall of the left ventricle shows focal subserosal lymphocytic infiltration (arrow) in the fibrotic lesion (H&E-6).



Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The right ventricle shows marked thinning of the myocardial layer (H&E-7).

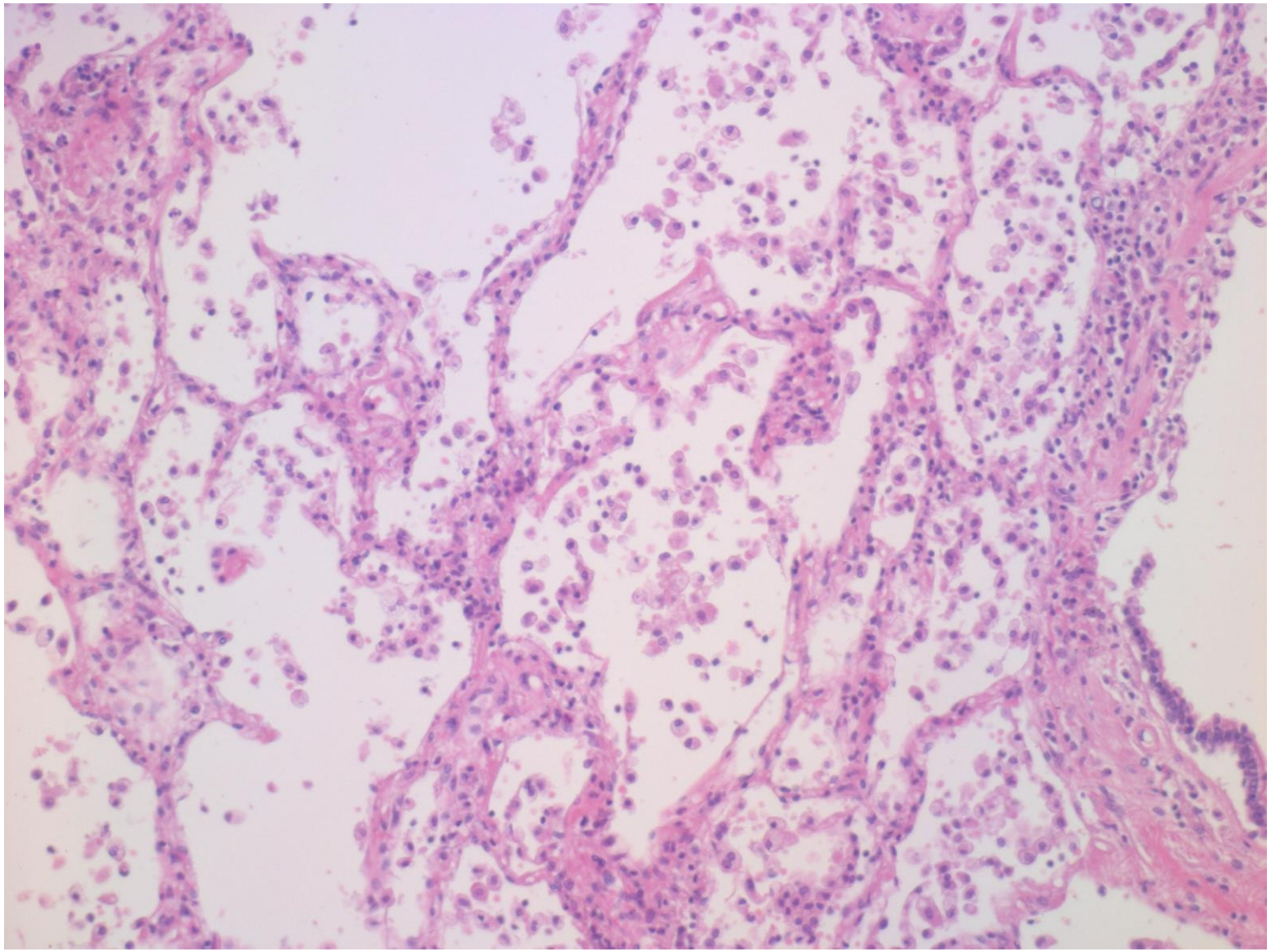


Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. Atherosclerotic calcification is seen in the coronary artery (left) and the aorta (right), as age-related changes (H&E-8).



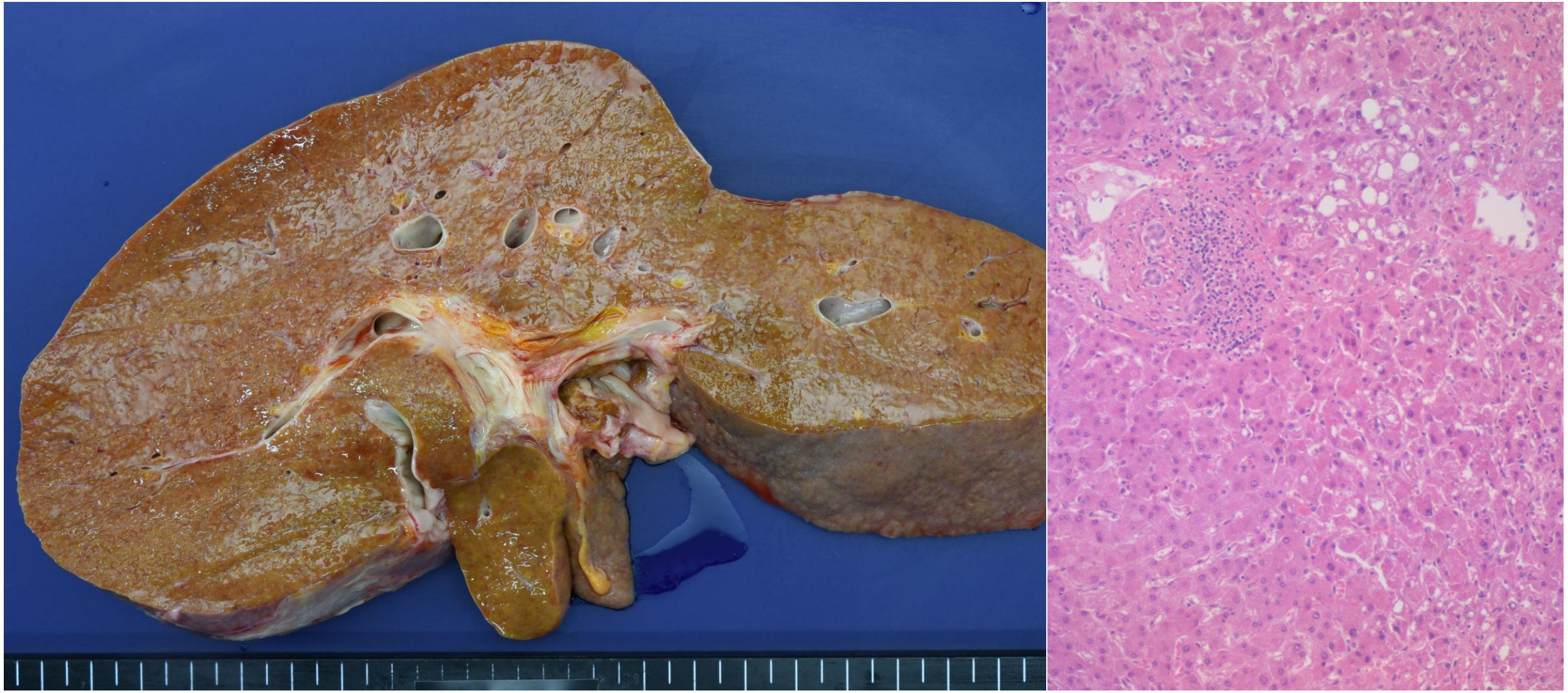
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The cut surfaces of the lung after formalin fixation reveal features of pulmonary edema (left: 350 g, right: 415 g)

肺（HE染色：肺胞内の心不全細胞）

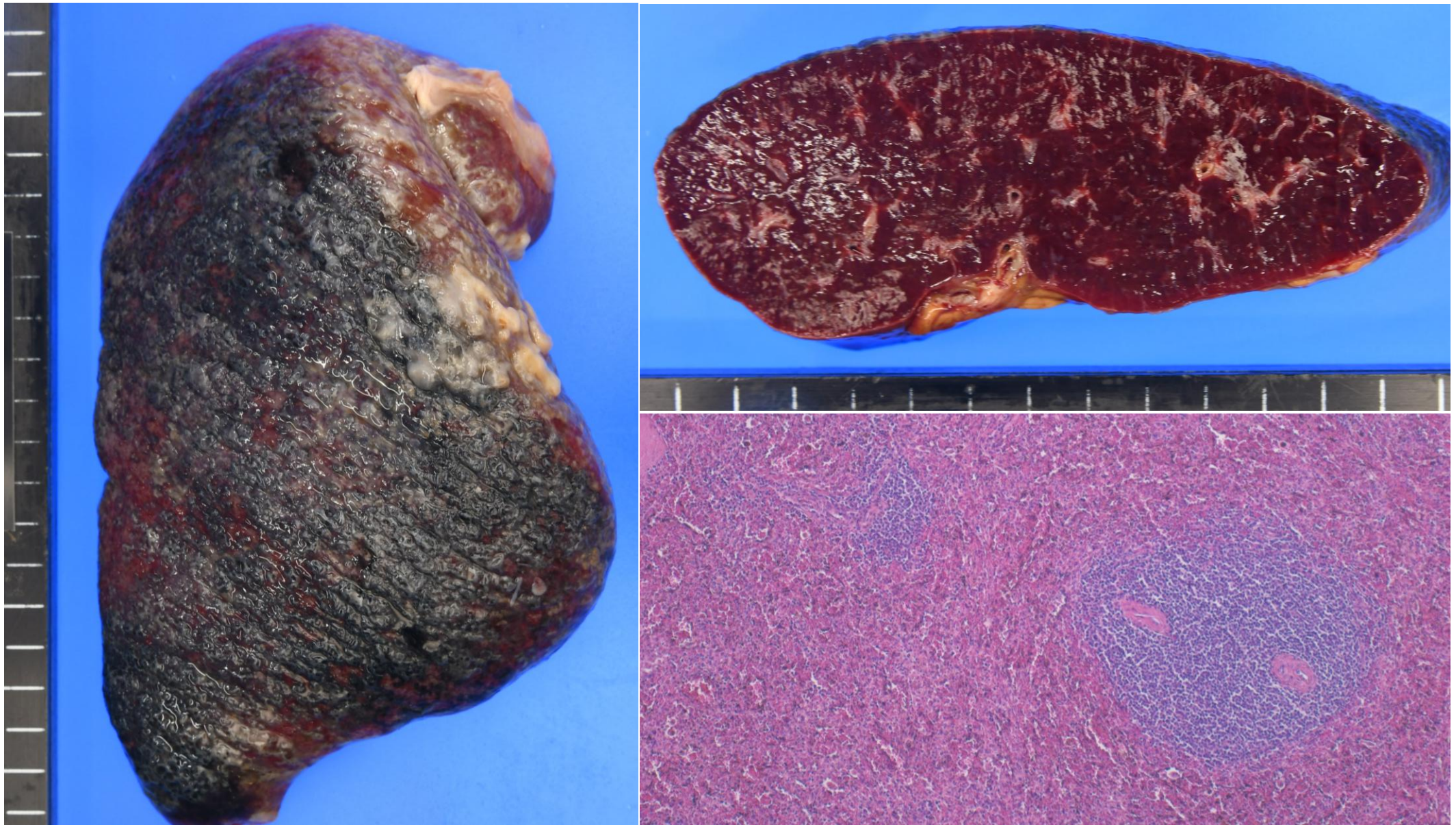


Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The alveolar lumina of the lung contain heart failure cells (H&E-9).

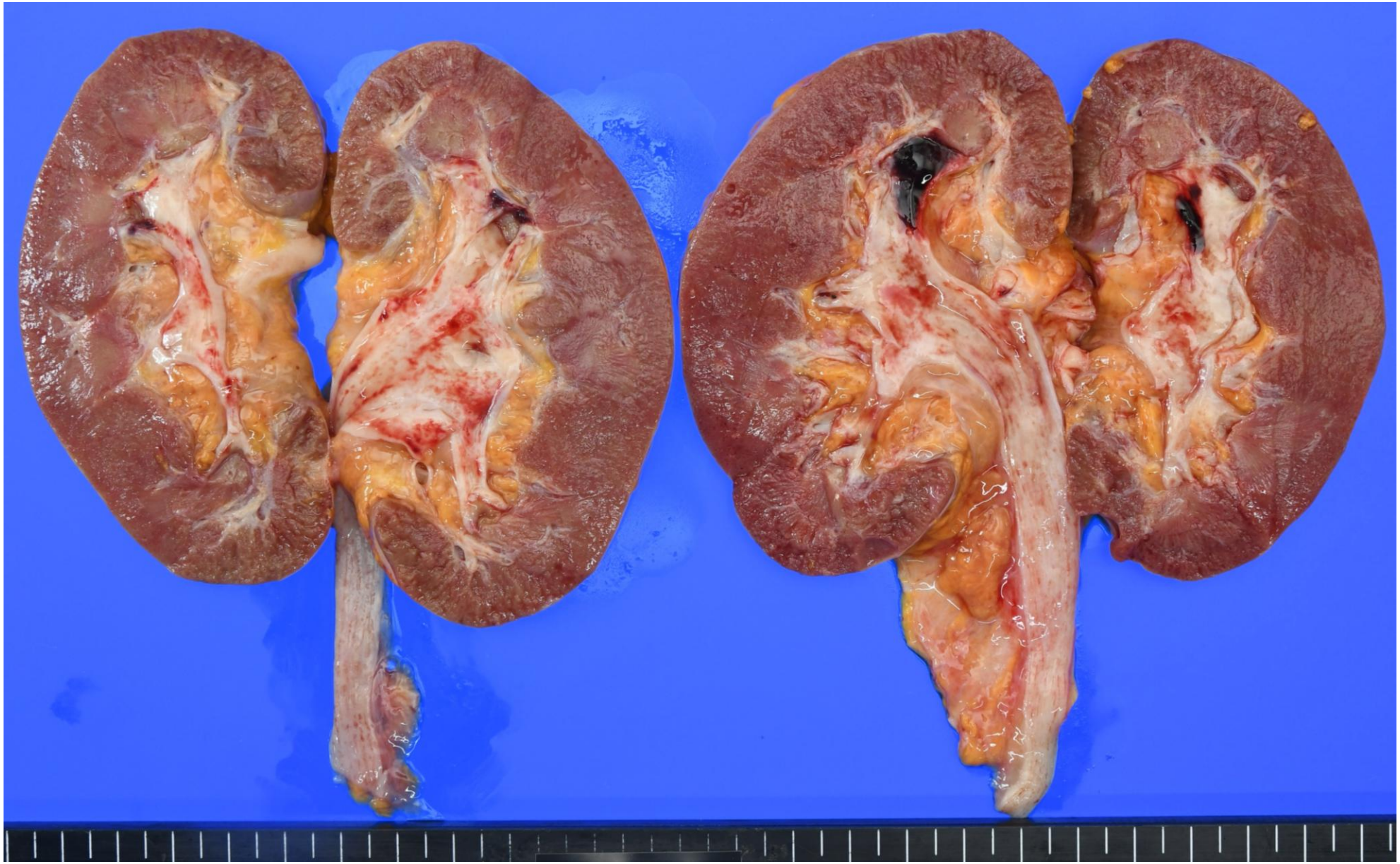
肝臓（ホルマリン固定後の割面像とHE染色、軽度のうっ血）



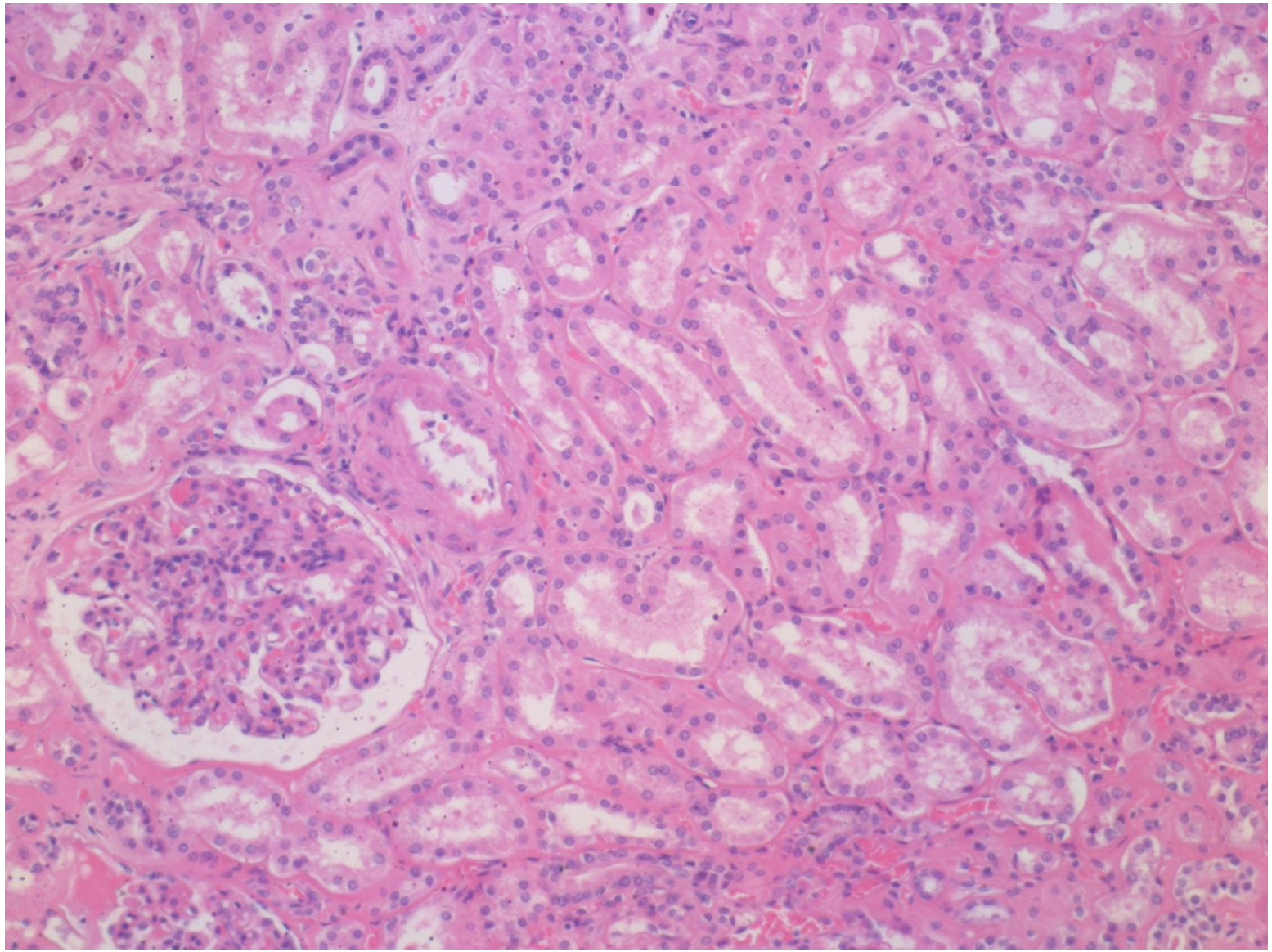
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The liver shows mild central congestion (left: gross appearance, right: H&E-10).



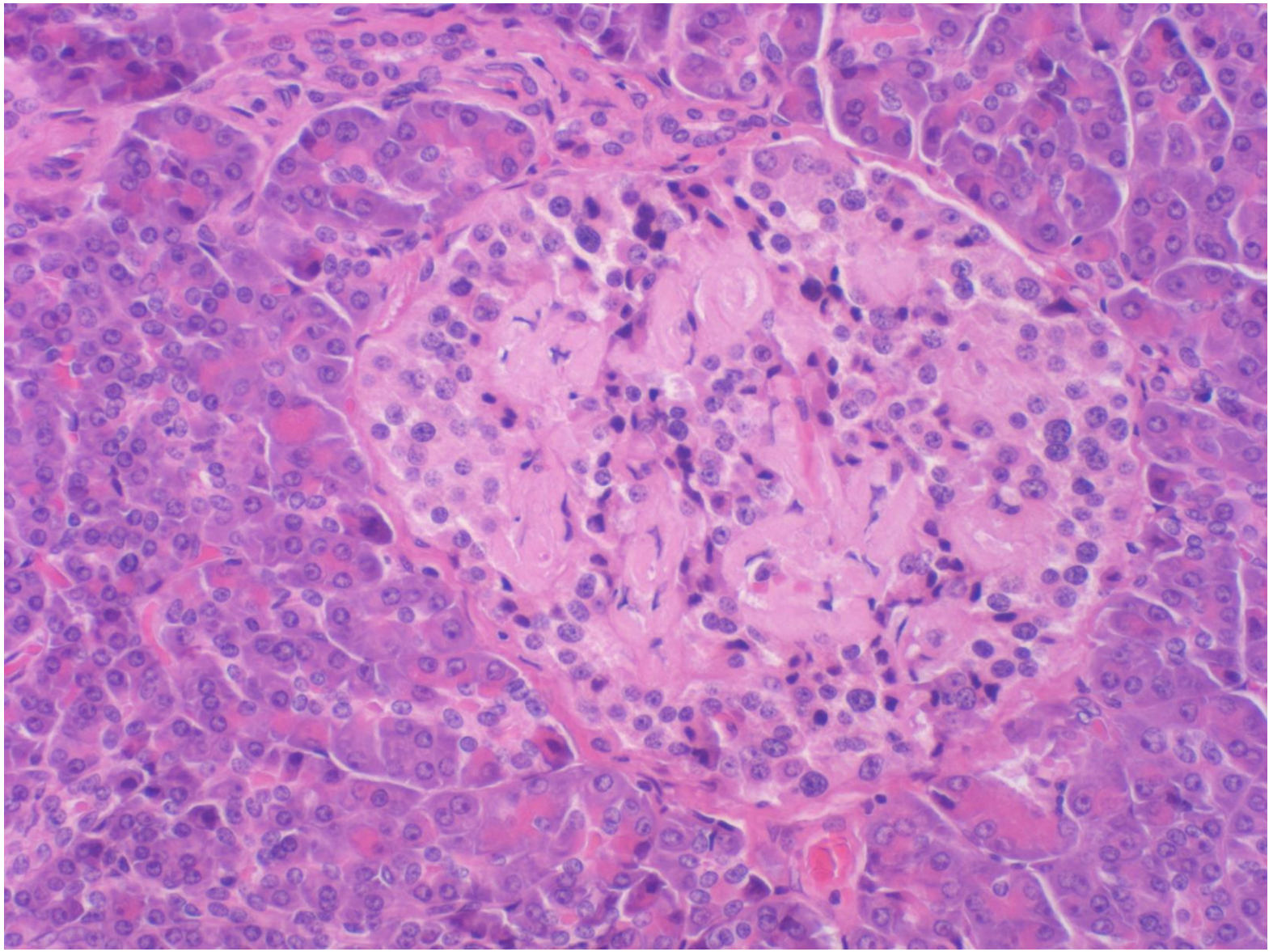
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The spleen (220 g) shows chronic congestion with a sugar-coated capsule (left: gross appearance, right top: cut surface, right bottom: H&E-11).



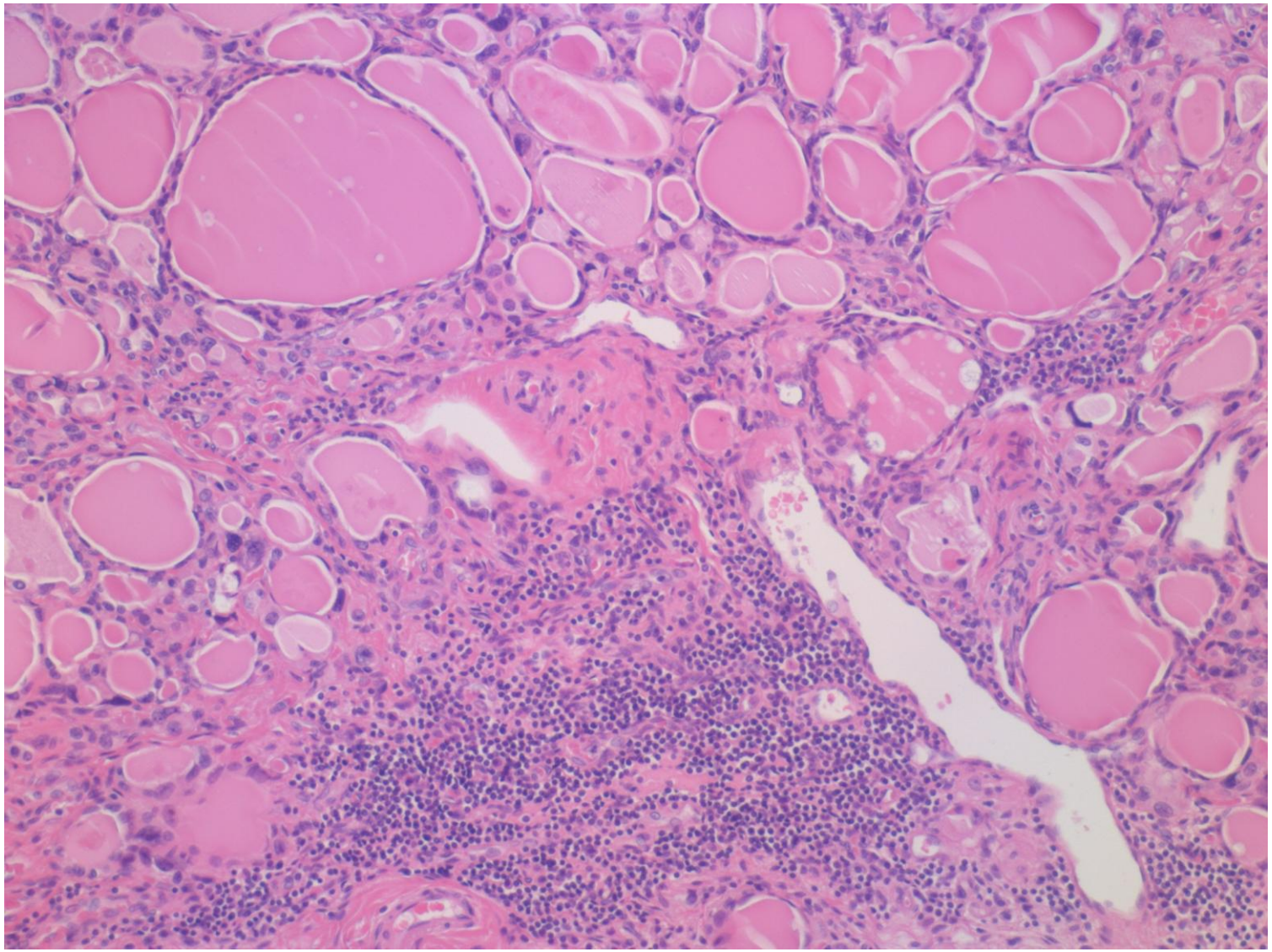
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The kidneys (left: 125 g, right: 115 g) show mild swelling and congestion.



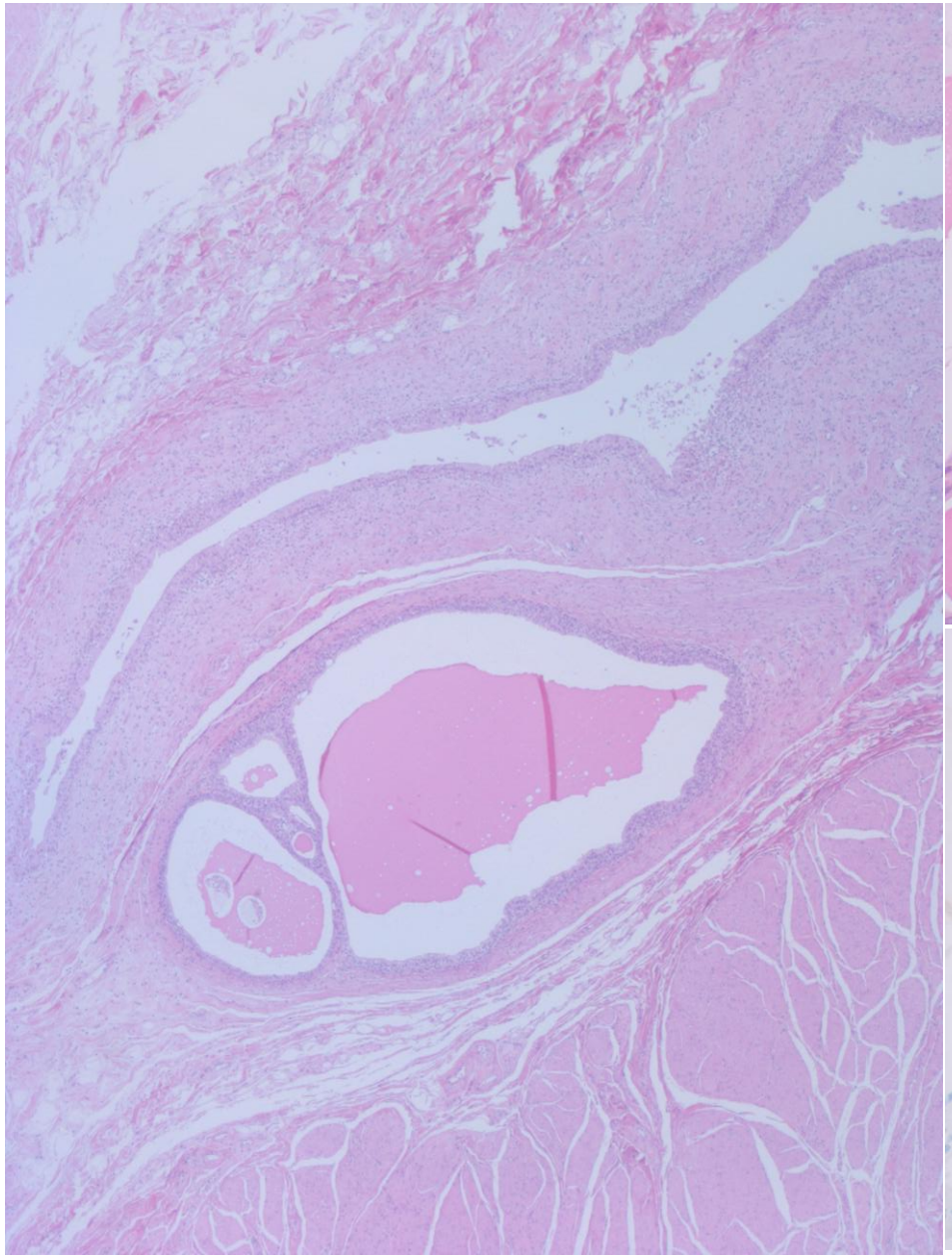
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The kidney microscopically shows an early stage of acute tubular necrosis (H&E-12).

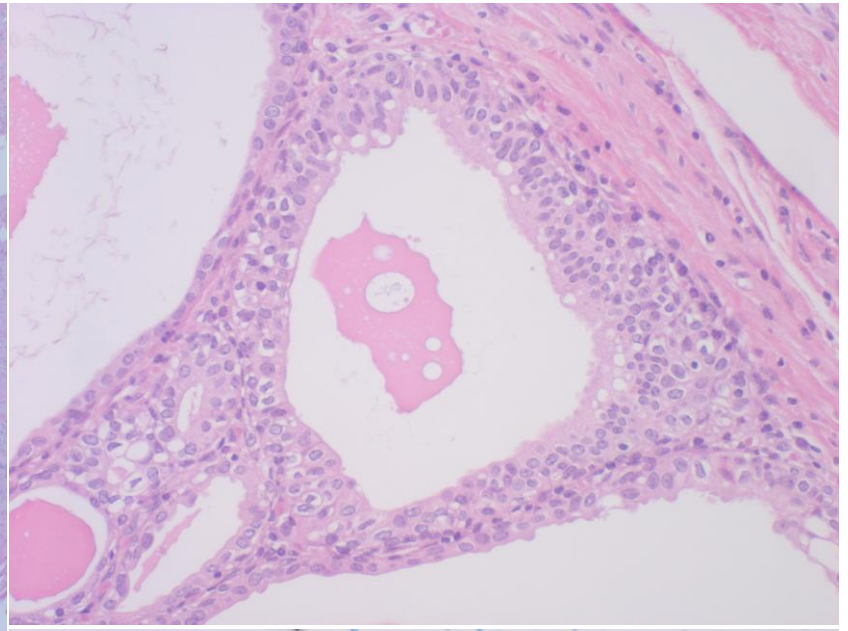


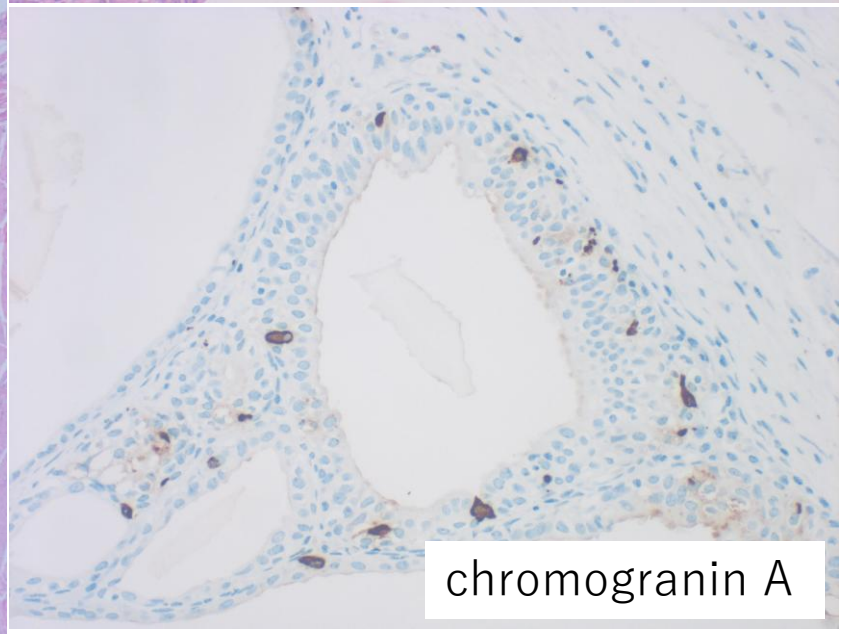
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The pancreatic islet microscopically shows amyloid deposition, strongly suggesting the complication of diabetes mellitus (H&E-13).



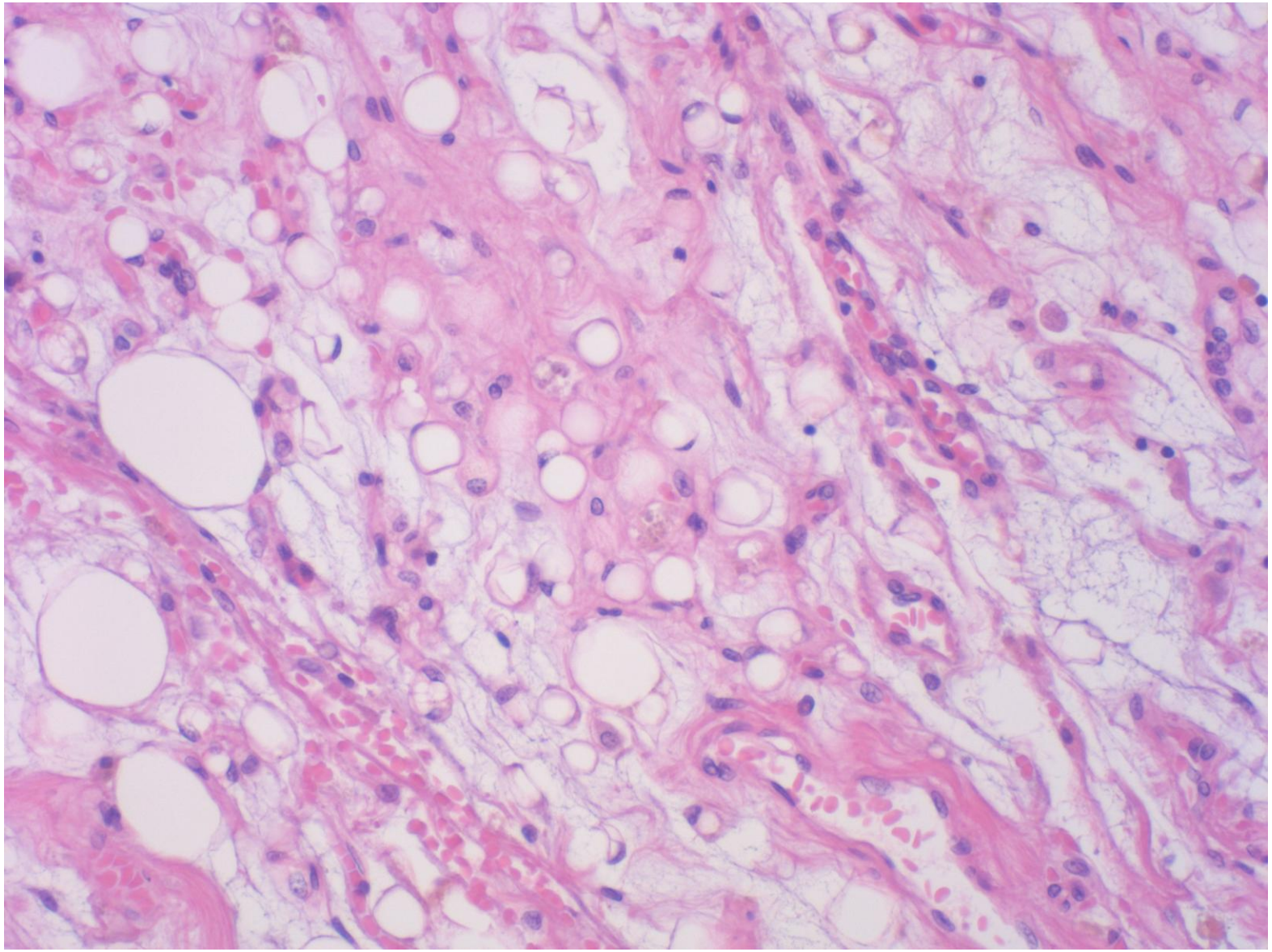
Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The thyroid gland (11 g) shows microscopic features of chronic lymphocytic thyroiditis (H&E-14).







Patent urachus at the tip of the urinary bladder. Physiological remnant of the urachal ducts contain a good number of neuroendocrine cells (H&E and immunostaining for chromogranin A).



Familial dilated cardiomyopathy seen in a 75 y-o male patient with an intimate genetic background. The patient died of chronic heart failure. The periadrenal fat tissue shows serous atrophy of fat cells (H&E-15).

Final anatomical diagnoses (72 y-o male poatient)

1. Dilated cardiomyopathy, familial (heart weight: 480 g)
   a) left ventricular dilatation with marked free wall thinning
   b) myocardial fibrosis (patchy, with predominance in the posterior wall)
   c) right ventricular and atrial dilatation
   d) intimate family history of cardiomyopathy
2. Pulmonary edema (left: 350 g, right: 415 g)
   a) Subpleural hemorrhage (right lower lobe, focal)
   b) Heart failure cells in the lung
3. Systemic congestion (liver, kidneys, spleen and digestive tract)
4. Anasarca
   (ascites 4,200 mL, pleural effusion left 300 mL, righty: 250 mL, pericardial effution 320 mL)
   a) Sugar-coated spleen
5. Acute tubular necrosis (early stage, kidney weight left: 125 g, right: 115 g)
6. Amyloid deposition in Langerhans islets (focal)
7. Prostatic hyperplasia (mild)
8. Chronic lymphocytic thyroiditis (thyroid: 11 g)
9. Chronic bronchiolitis (mild)
10. Testicular atrophy (testis left: 21 g, right: 22 g)
11. Atherosclerosis of the aorta (moderate)
12. Patent urachus (urinary bladder apex, physiological)
13. Serous atrophy of fat tissue

Direct cause of death: Chronic heart failure