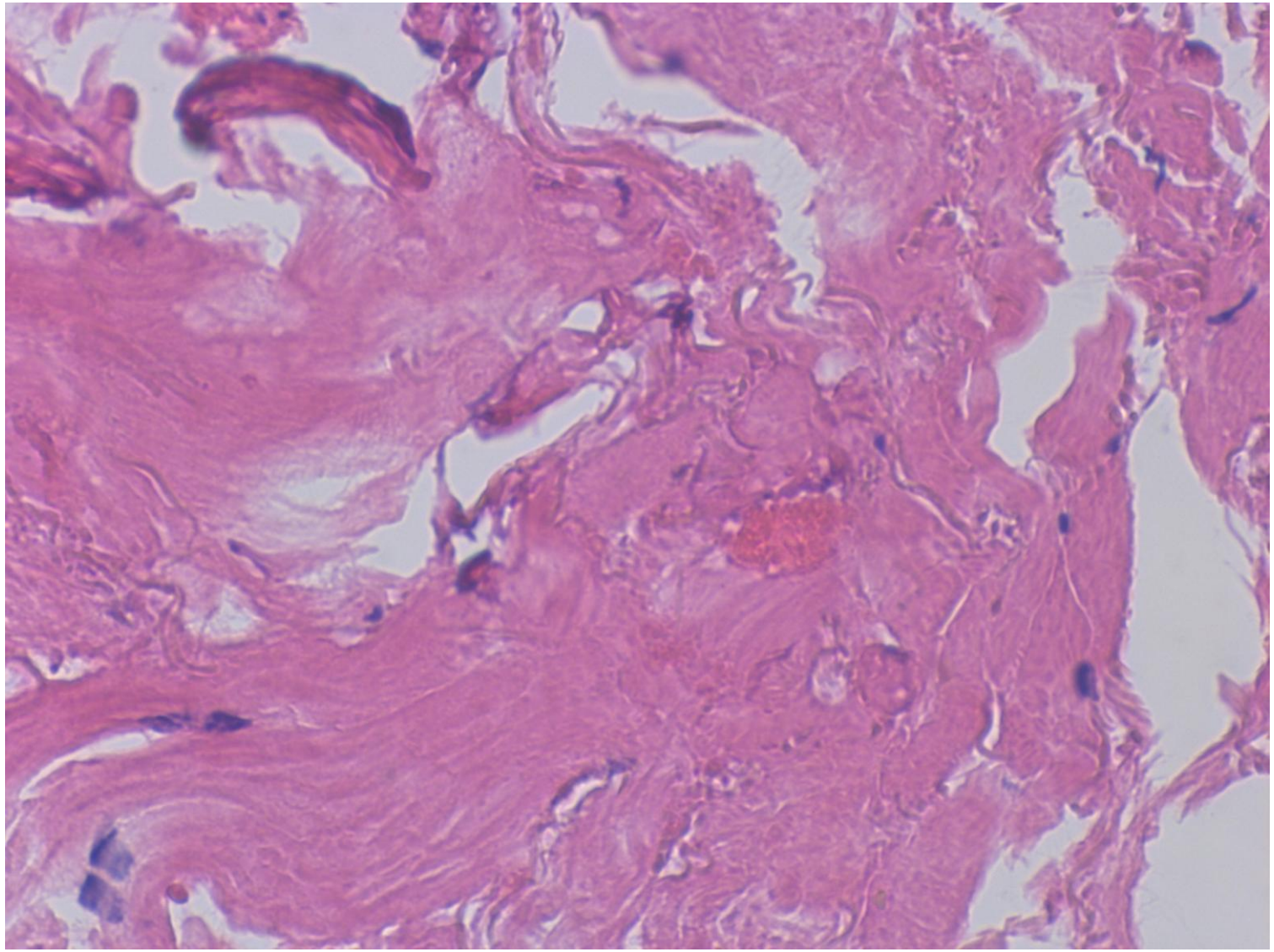
Subcutaneous biopsy for the diagnosis of cardiac amyloidosis

A reliable diagnosis of amyloidosis is based on a tissue biopsy. An exact and valid diagnosis including determination of amyloid typing is imperative. Biopsy has been directed towards a symptomatic organ such as the heart, liver and kidney. Screening biopsy is also useful for making a diagnosis of systemic amyloidosis. The sites of screening biopsy include the subcutaneous adipose tissue, gastrointestinal tract (the rectum, stomach or duodenum), lip (minor salivary glands), and bone marrow. Here presented is the usefulness of subcutaneous biopsy for the diagnosis of cardiac amyloidosis predominantly caused by AL type and transthyretin (ATTR) type. Congo red staining can detect the amyloid deposition. It should be noted that Congo red may give false positive staining in collagen fibers. The importance of electron microscopic analysis should also be emphasized.

Ref.-1: Benson MD, et al. Tissue biopsy for the diagnosis of amyloidosis: experience from some centres. Amyloid 2021; 29(1): 8-13. doi: 10.1080/13506129.2021.1994386

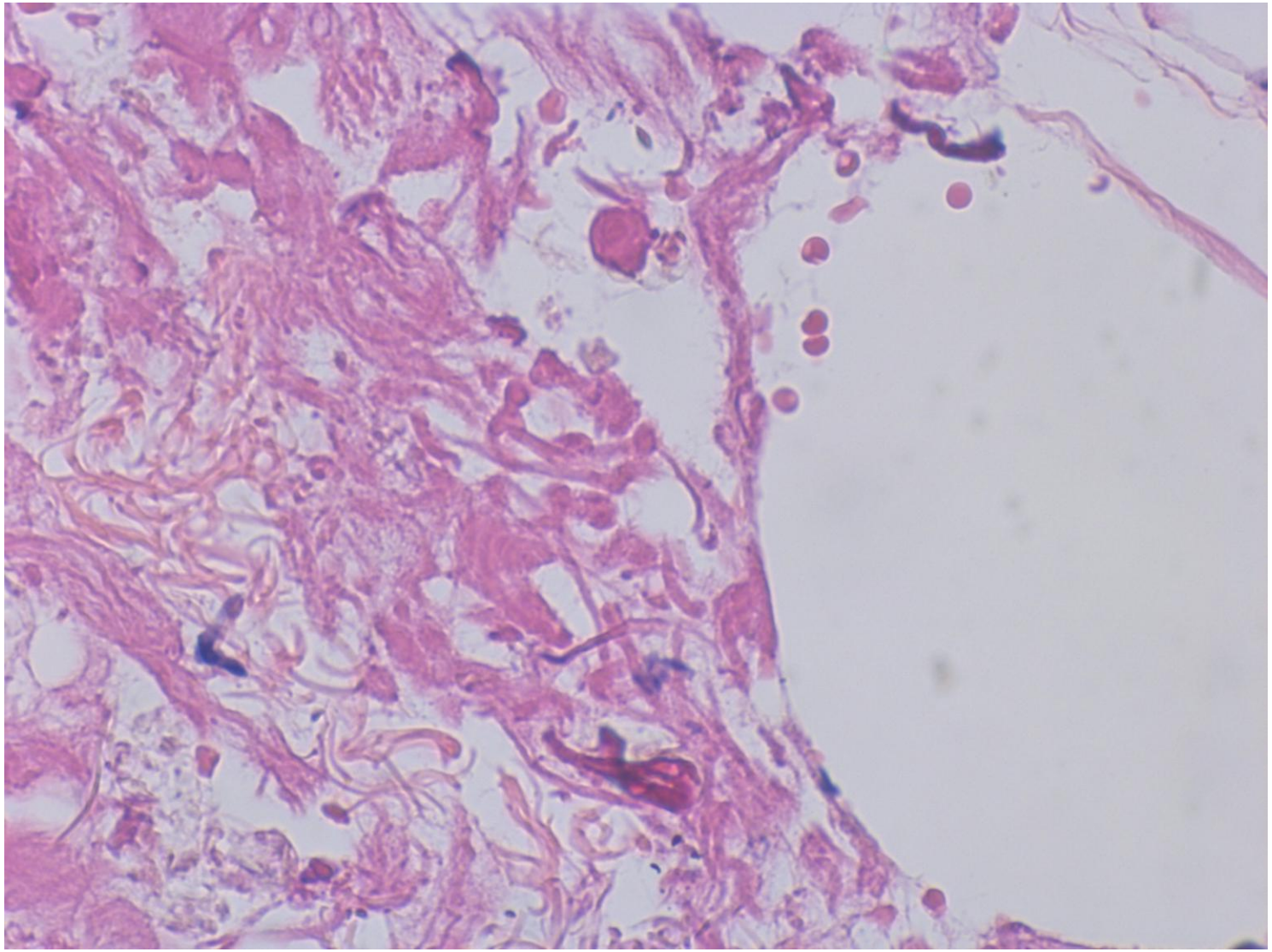
Ref.-2: Movila DE, et al. Cardiac amyloidosis: a narrative review of diagnostic advances and emerging therapies. Biomedicines 2025; 13(5): 1230. doi: 10.3390/biomedicines13051230

Case 1
84F
amyloid+



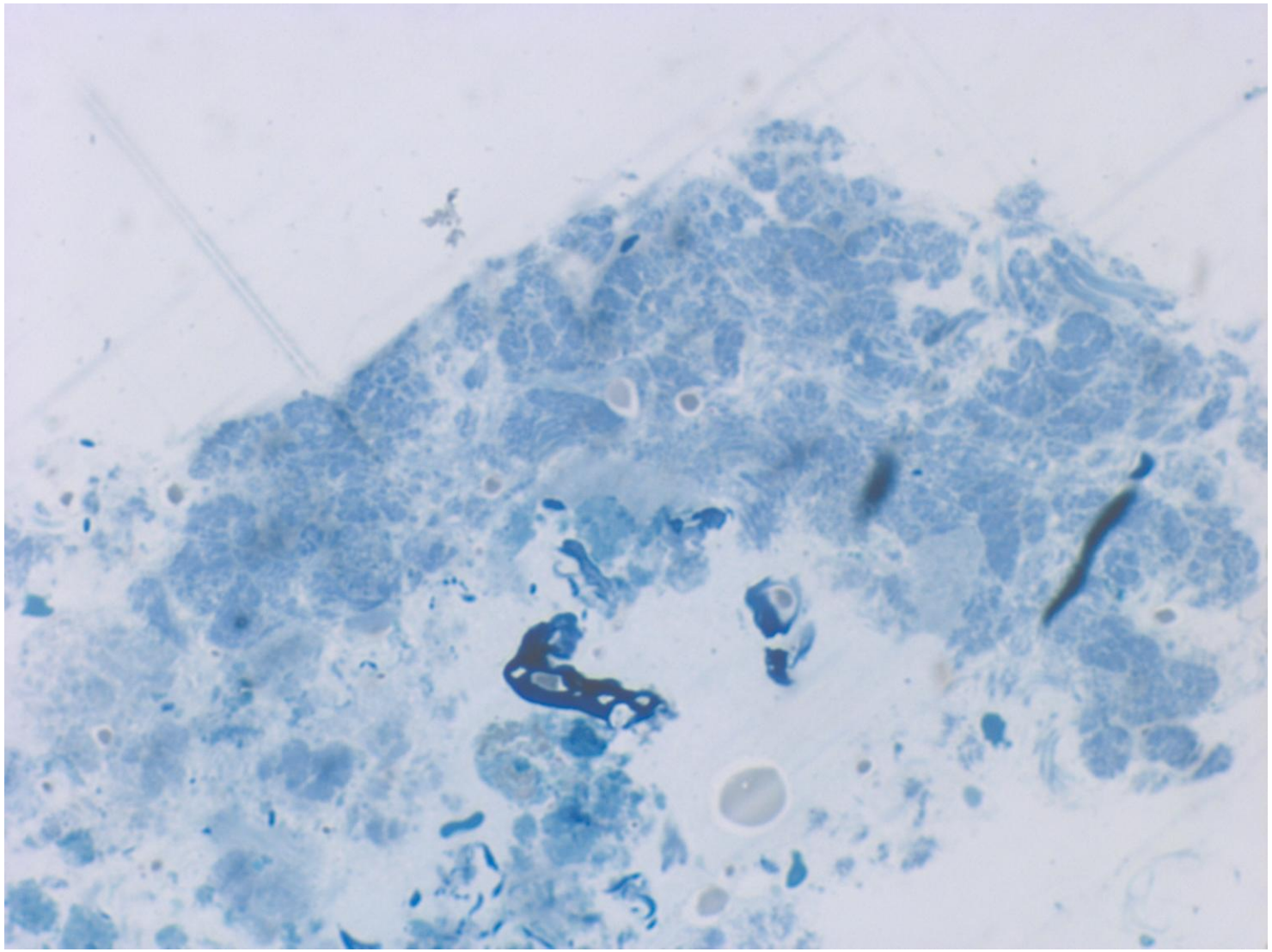
An 84 y-o female patient manifested bradycardiac atrial fibrillation. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the chest wall was biopsied when a pacemaker was implanted. Collagenous component among the subcutaneous fat tissue has eosinophilic globular condensations (H&E-1).

Case 1
84F
amyloid+



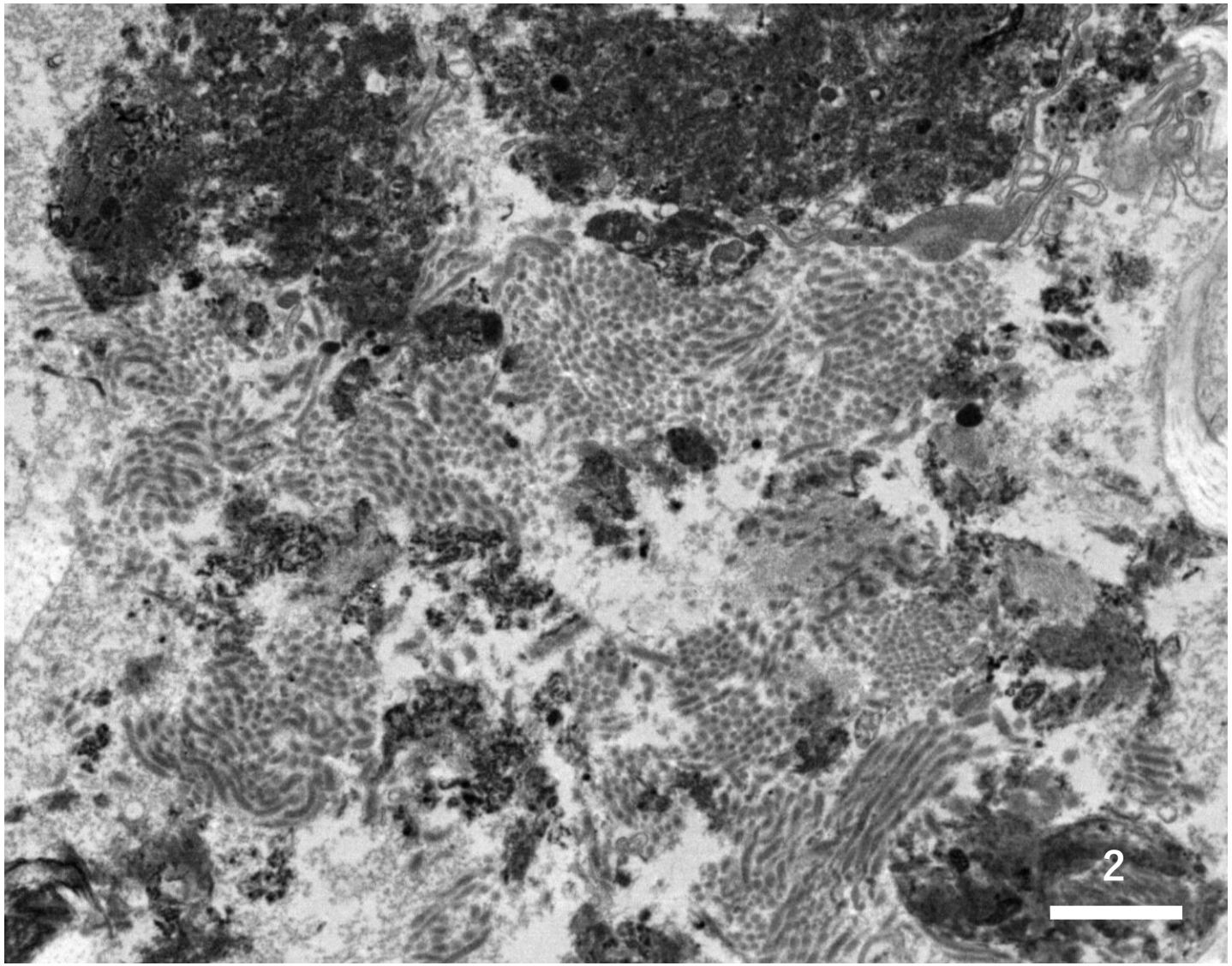
An 84 y-o female patient manifested bradycardiac atrial fibrillation. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the chest wall was biopsied when a pacemaker was implanted. Eosinophilic globular condensations are seen among a collagenous component adjacent to the subcutaneous fat tissue (H&E-2).

Case 1
84F
amyloid+



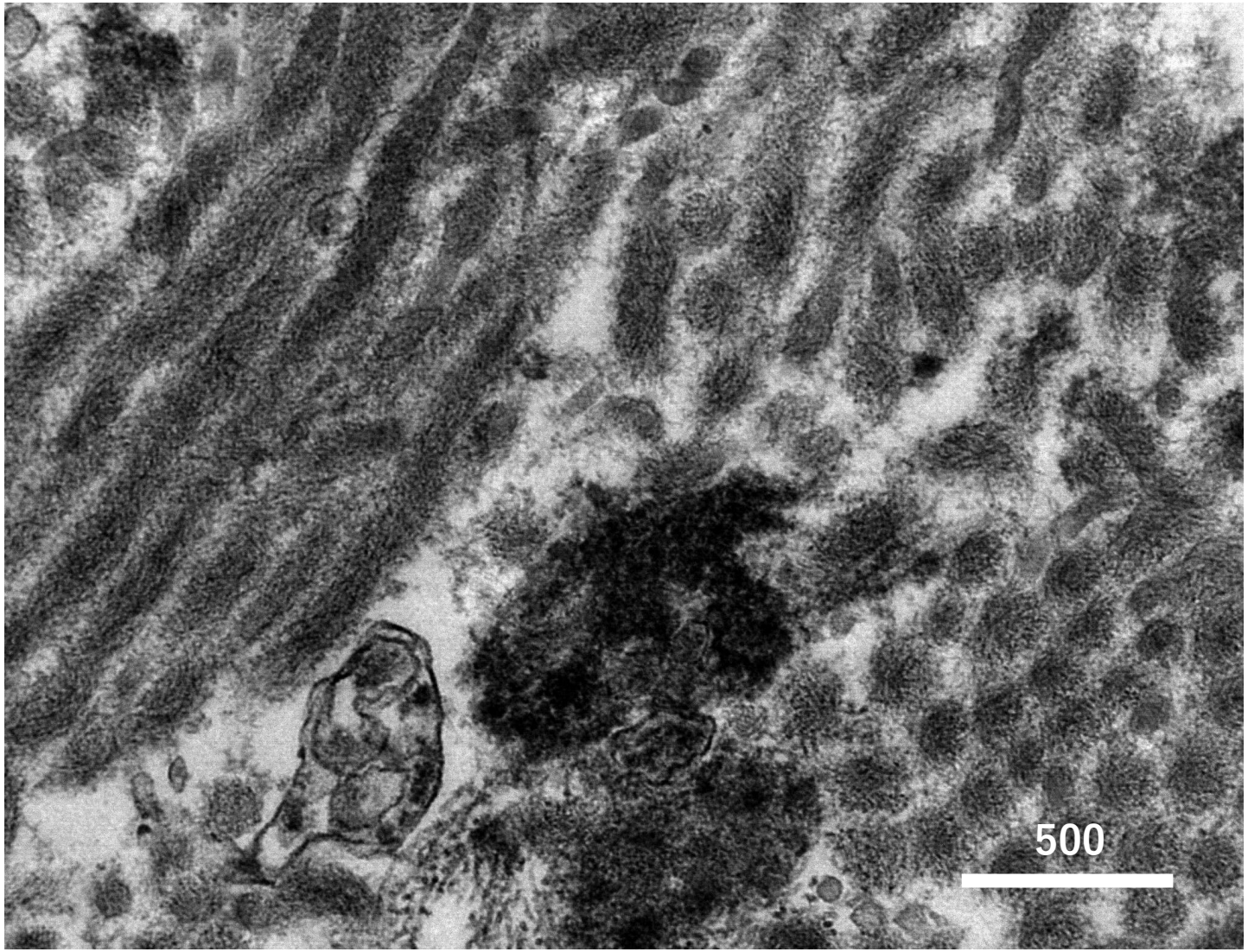
An 84 y-o female patient manifested bradycardiac atrial fibrillation. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the chest wall was biopsied when a pacemaker was implanted. Globular condensations are seen among a collagenous component adjacent to the subcutaneous fat tissue (Toluidine blue-stained thick section for EM study).

Case 1
84F
amyloid+



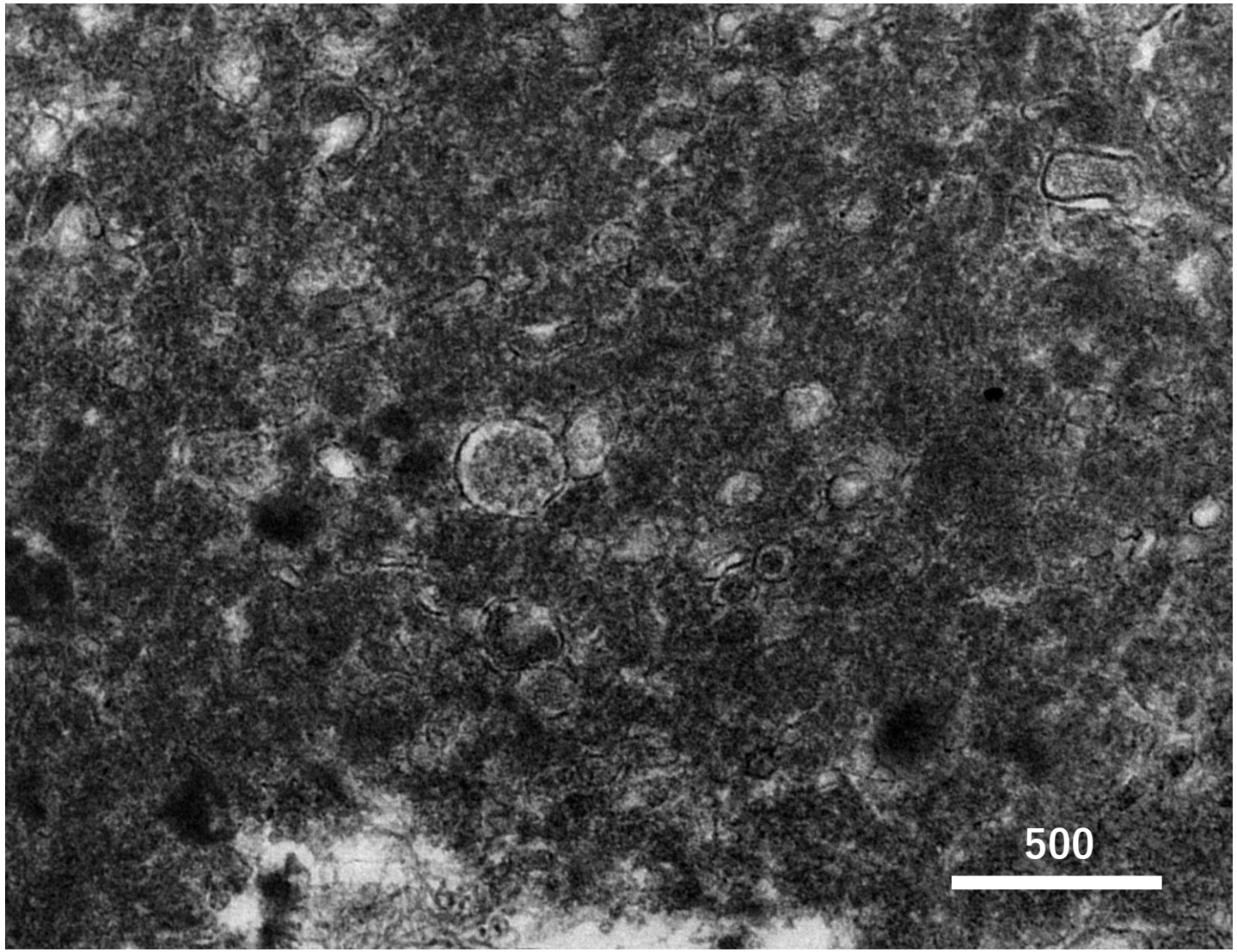
An 84 y-o female patient manifested bradycardiac atrial fibrillation. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the chest wall was biopsied when a pacemaker was implanted. Ultrastructurally, multifocal clusters of electron-dense fibrillar material are observed among the collagen fibers, corresponding to the eosinophilic globular material (TEM-1).

Case 1
84F
amyloid+



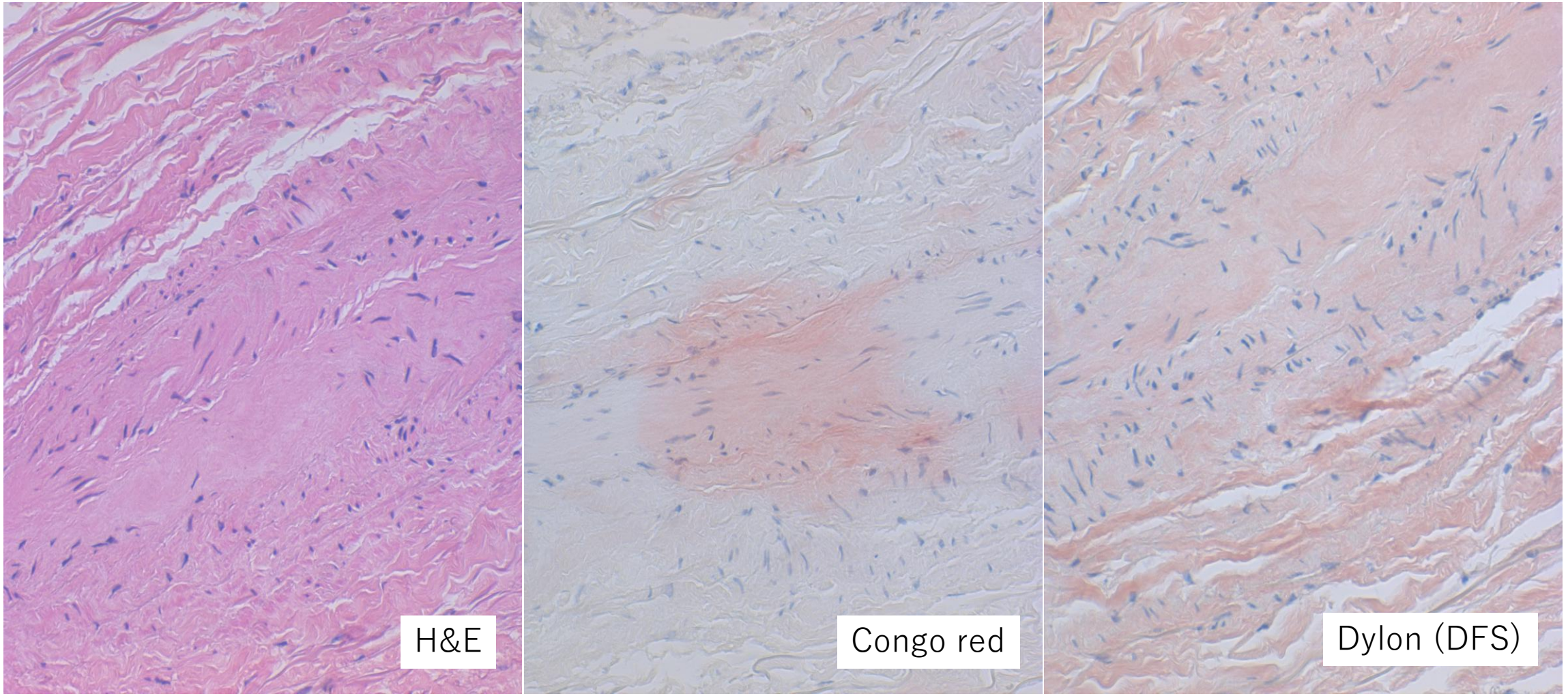
An 84 y-o female patient manifested bradycardiac atrial fibrillation. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the chest wall was biopsied when a pacemaker was implanted. Ultrastructurally, a cluster of electron-dense fibrillar material is observed among the collagen fibers, corresponding to the eosinophilic globular material (TEM-2).

Case 1
84F
amyloid+



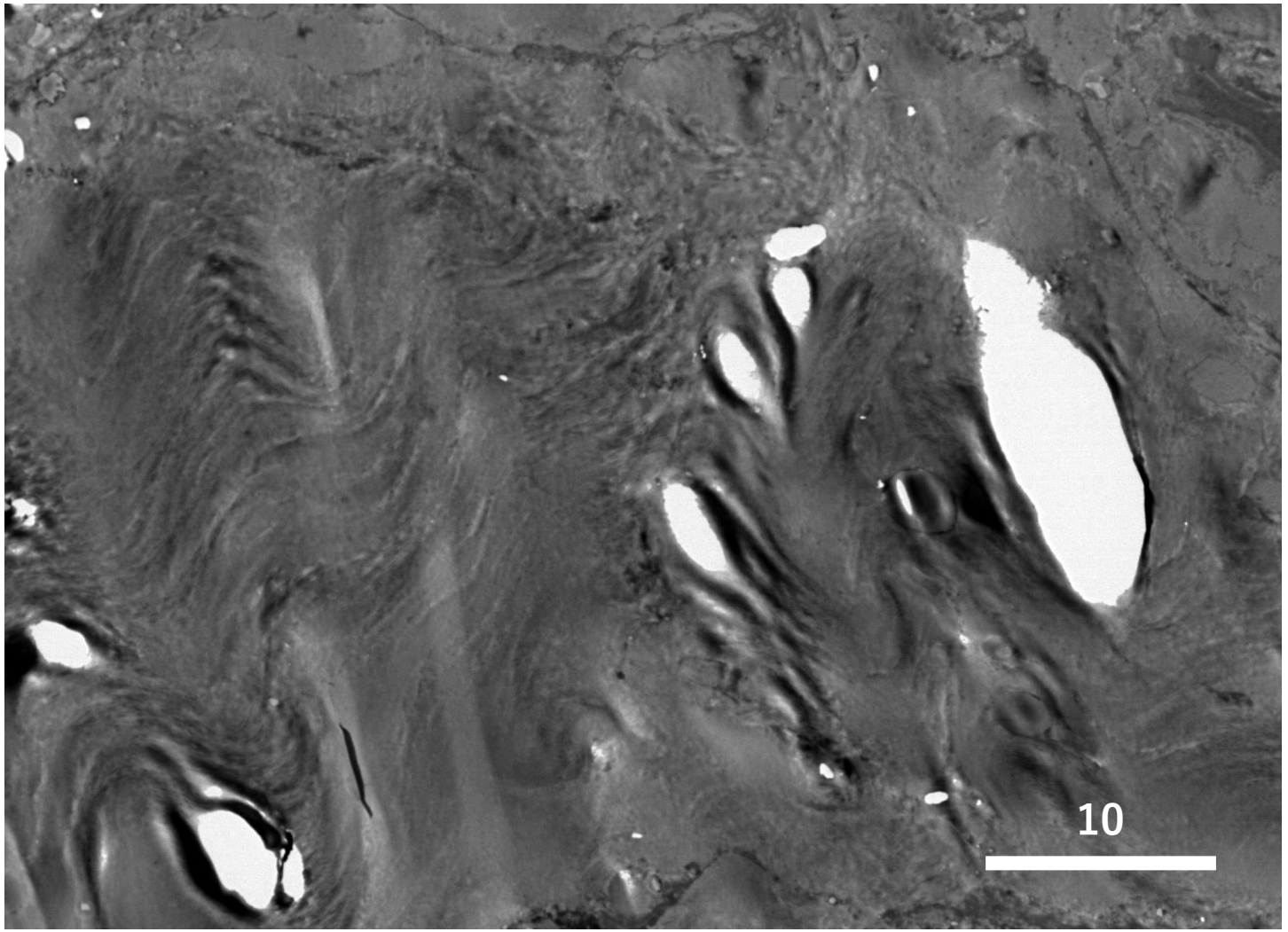
An 84 y-o female patient manifested bradycardiac atrial fibrillation. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the chest wall was biopsied when a pacemaker was implanted. Ultrastructurally, the electron-dense non-branching fibrillar material is consistent with amyloid fibrils (TEM-3).

Case 2, 83F, amyloid-



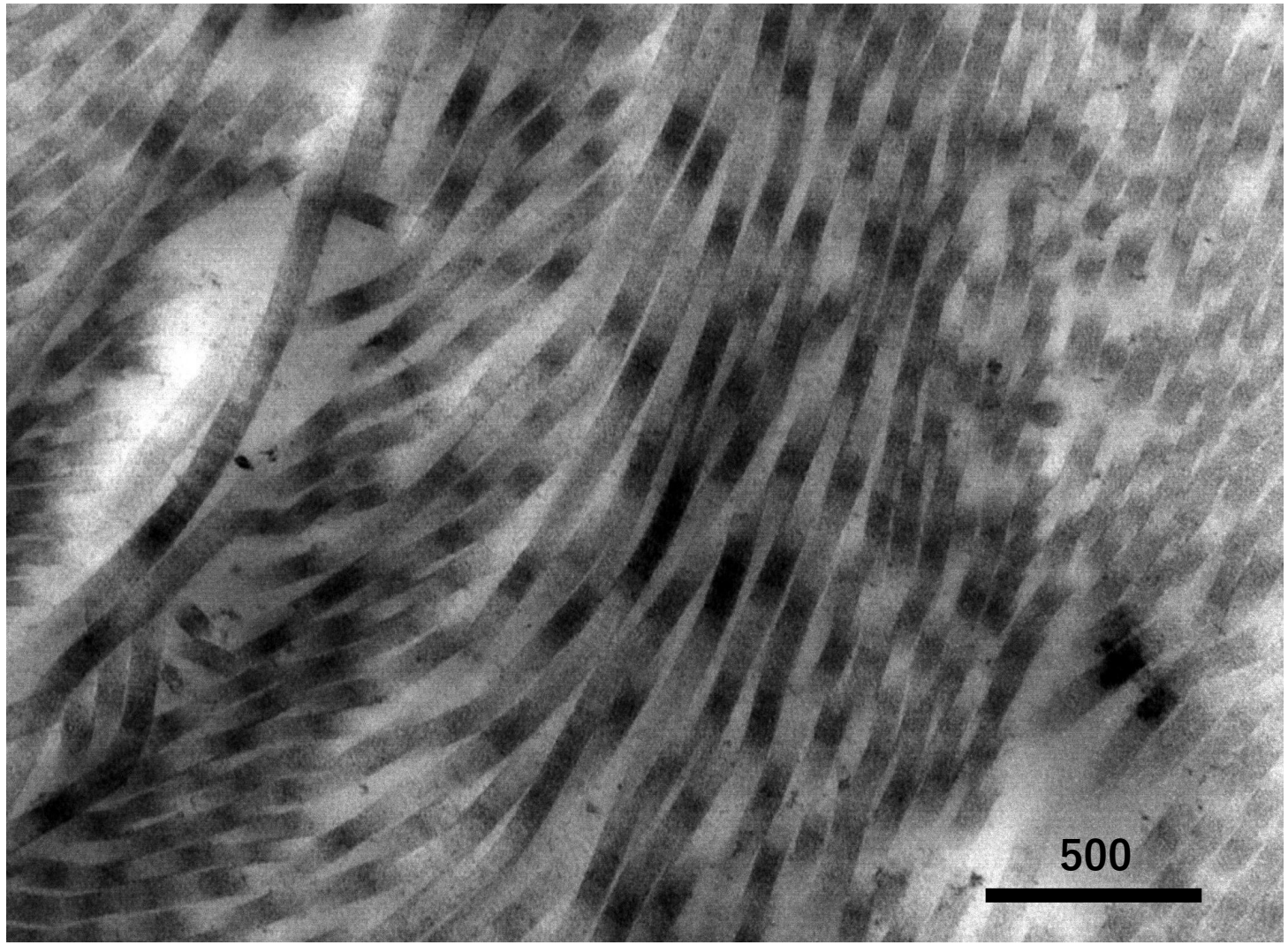
An 83 y-o female patient manifested cardiac failure. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the thigh was biopsied. The collagen bundles were included in the subcutaneous tissue. Congo red appears to be positive, while Dylon staining discloses nonspecific staining on the collagen fibers. Attention should be paid for nonspecific reactivity of amyloid staining on the collagen fibers (left: H&E, center: Congo red, right: Dylon).

Case 2
83F
amyloid-



An 83 y-o female patient manifested cardiac failure. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the thigh was biopsied. The collagen bundles were included in the subcutaneous tissue. Ultrastructurally, no amyloid fibrils can be seen among the collagen fibers (TEM-a).

Case 2
83F
amyloid-



An 83 y-o female patient manifested cardiac failure. Cardiac amyloidosis was clinically suspected, and subcutaneous tissue of the thigh was biopsied. The collagen bundles were included in the subcutaneous tissue. Ultrastructurally, interlacing bundles of collagen fibers are noted but without amyloid fibrils (TEM-b).