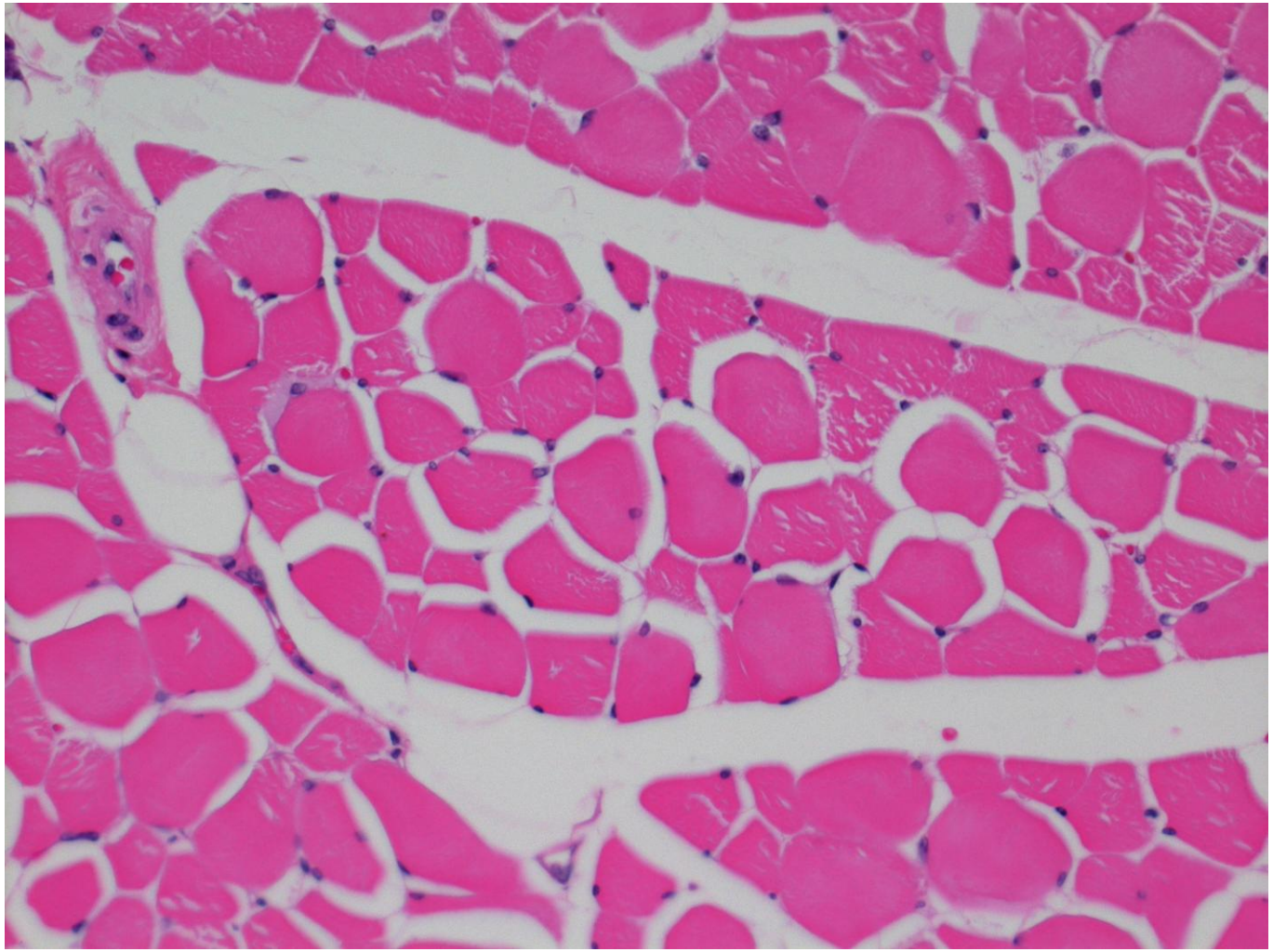
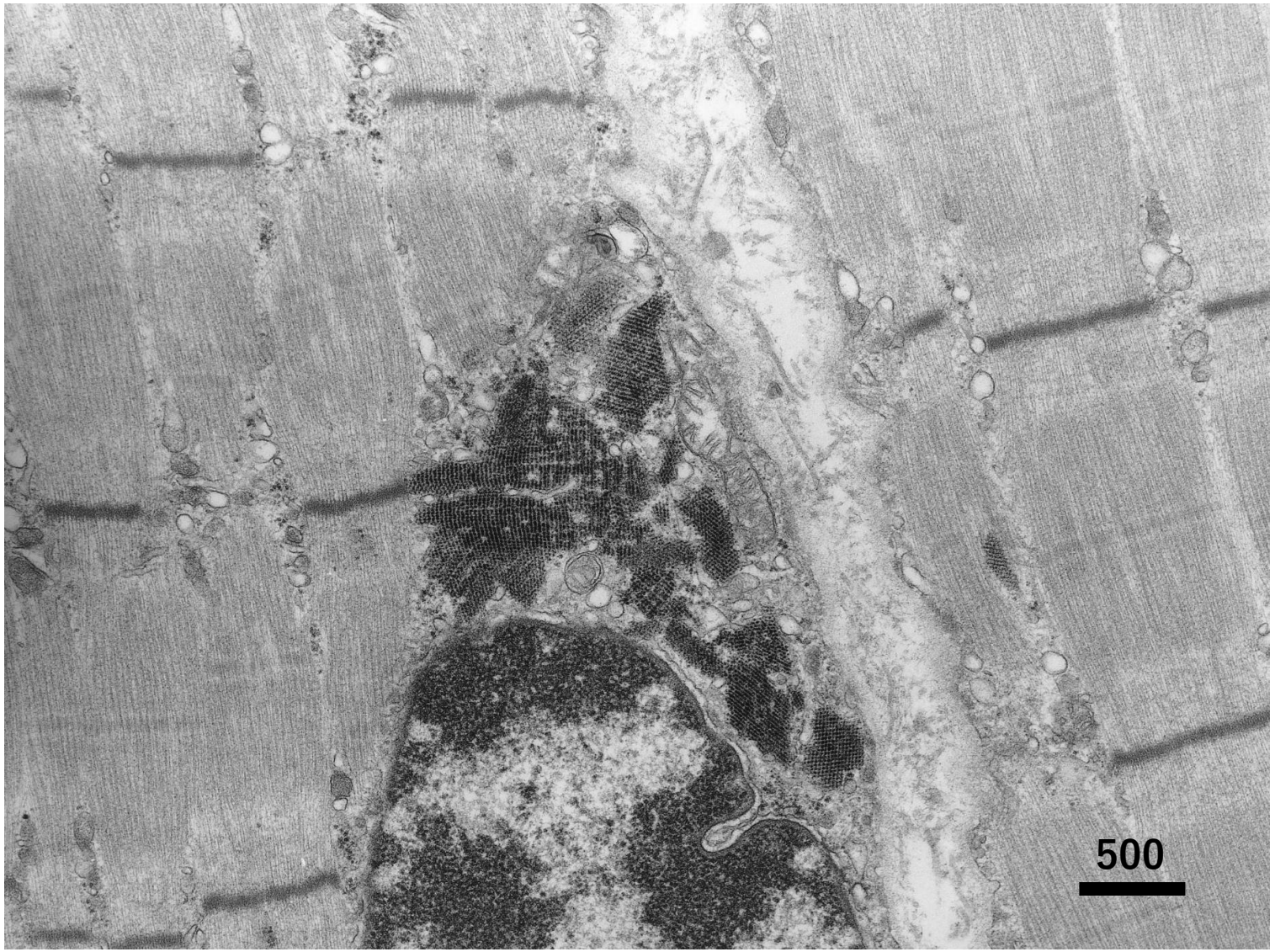
Nemaline myopathy

Nemaline myopathy (rod myopathy) is a hereditary neuromuscular disorder accompanying proximal muscle weakness, hypoventilation, swallowing dysfunction and impaired speech ability. The muscle exhaustion usually happens between ages 20–50. The severity of the symptoms varies and may change throughout life. Mutations of the NEB gene are transmitted with autosomal recessive trait. ACTA1 gene mutations cause nemaline myopathy in autosomal dominant manner. Microscopically, Gomori trichome stain shows red to purple colored, rod-shaped inclusions in the subsarcolemmal region of myofibers. Ultrastructurally, electron dense rod to ovoid shaped deposits, measuring 1-7 μm in length, are observed. The rods show continuity with Z-disks and exhibit a lattice structure. “Nemaline bodies” are immunoreactive for alpha-actinin. “Nema” is a Greek word meaning thread.

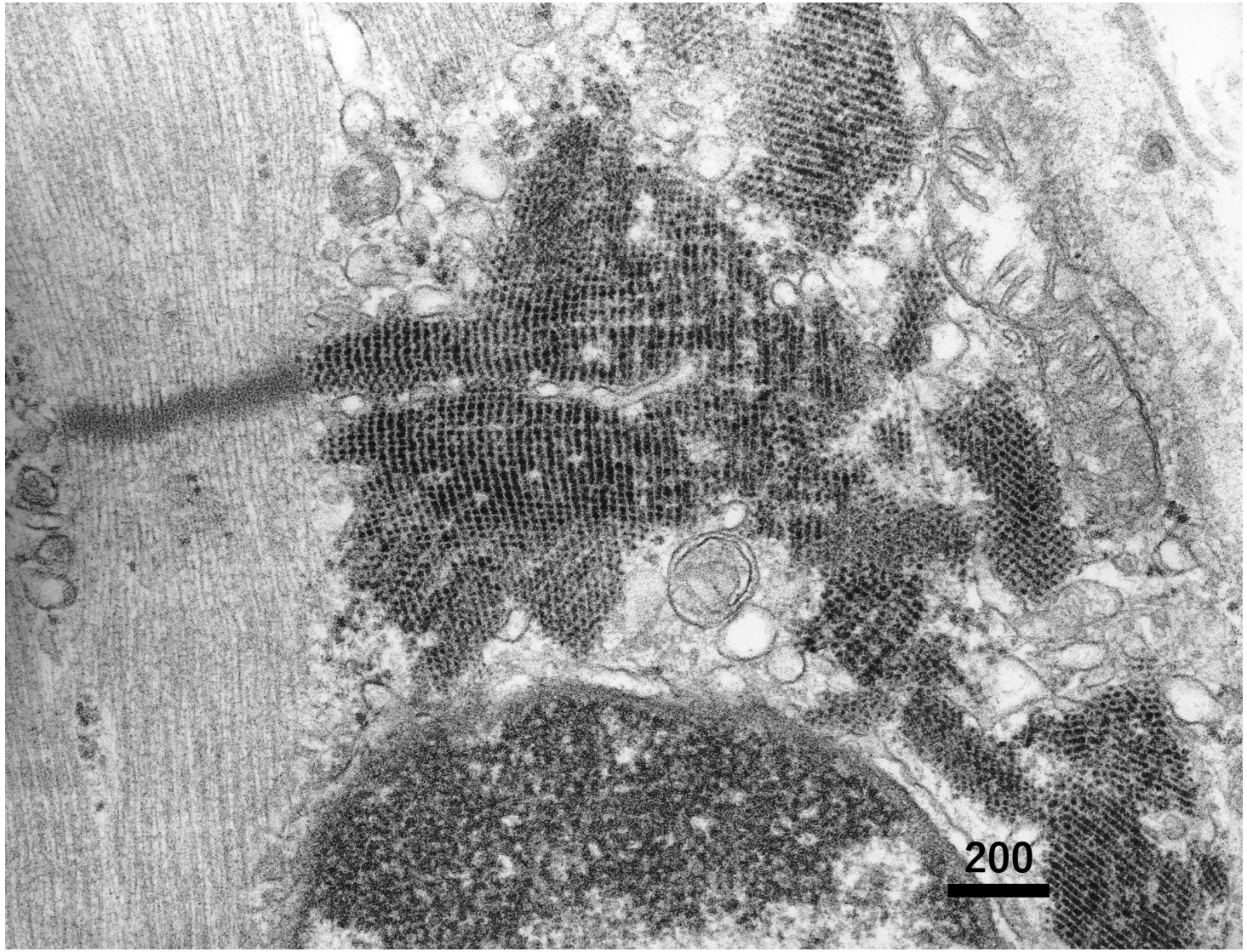
A 16-year-old boy suddenly died by heart failure. Autopsy revealed diffuse myocarditis, complicated with nemaline myopathy in the striated muscle. Cardiac disease in nemaline myopathy usually manifests as cardiomyopathy leading to heart failure. When respiratory muscles are affected, the right side of the heart may be involved secondarily. Myocarditis seen in the present case may be incidental.



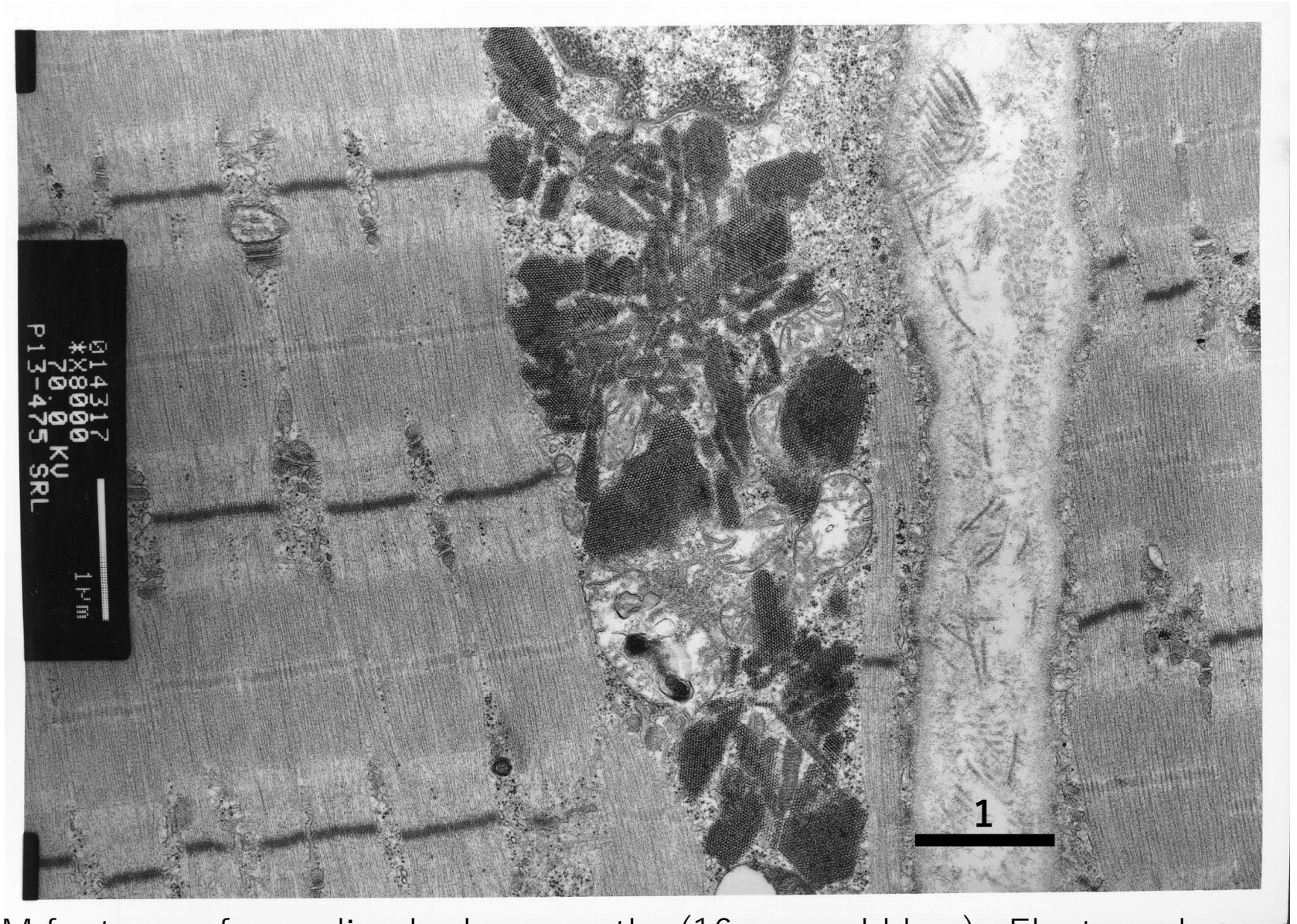
Nemaline body myopathy in a 16-year-old boy). Psoas muscle was sampled at autopsy. Minimal abnormality is discerned light microscopically. H&E



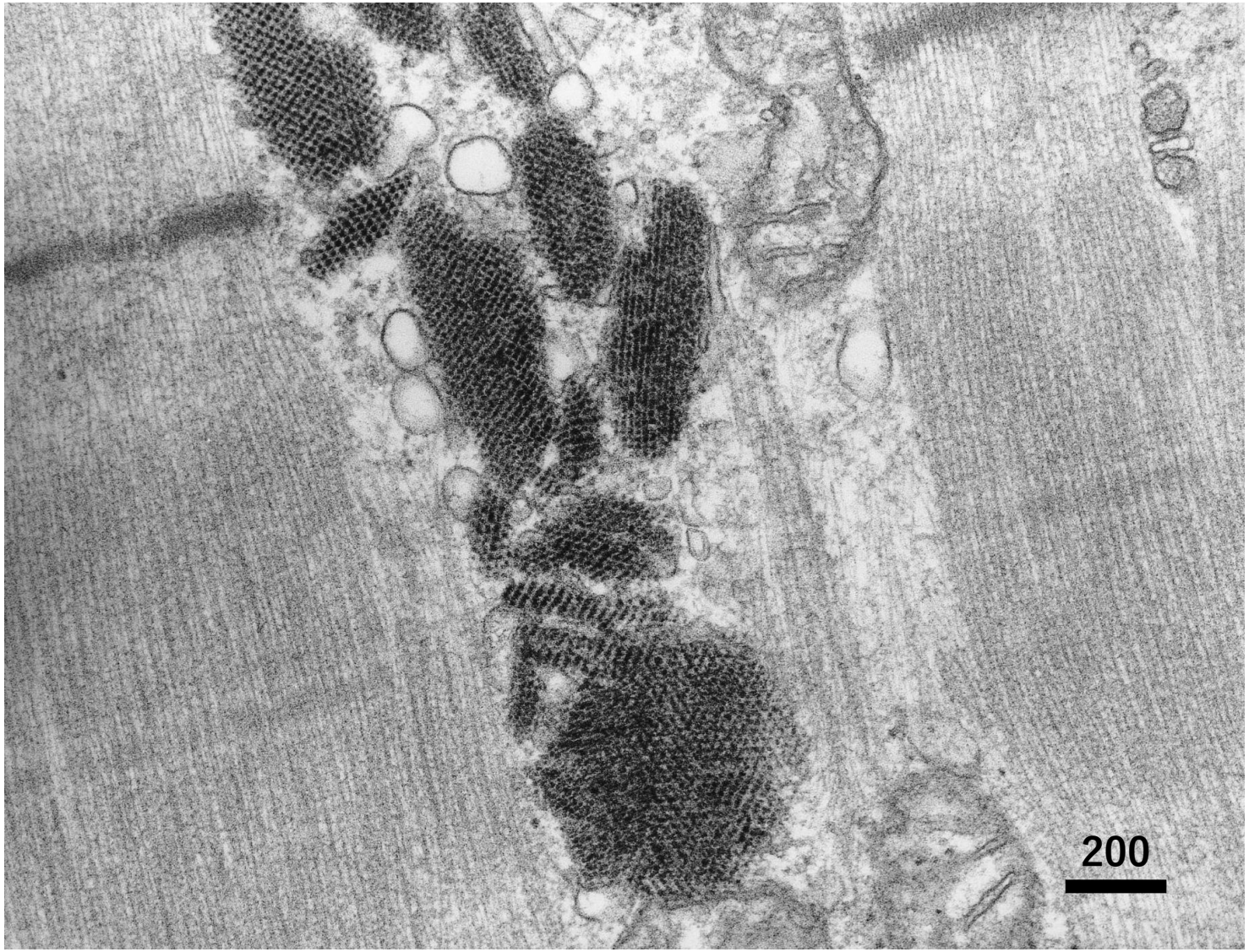
EM features of nemaline body myopathy (16-year-old boy). Electron-dense, lattice-like inclusion bodies are located beneath the subsarcolemmal region. Note the direct continuity of the inclusion bodies to the Z-band. EM-1



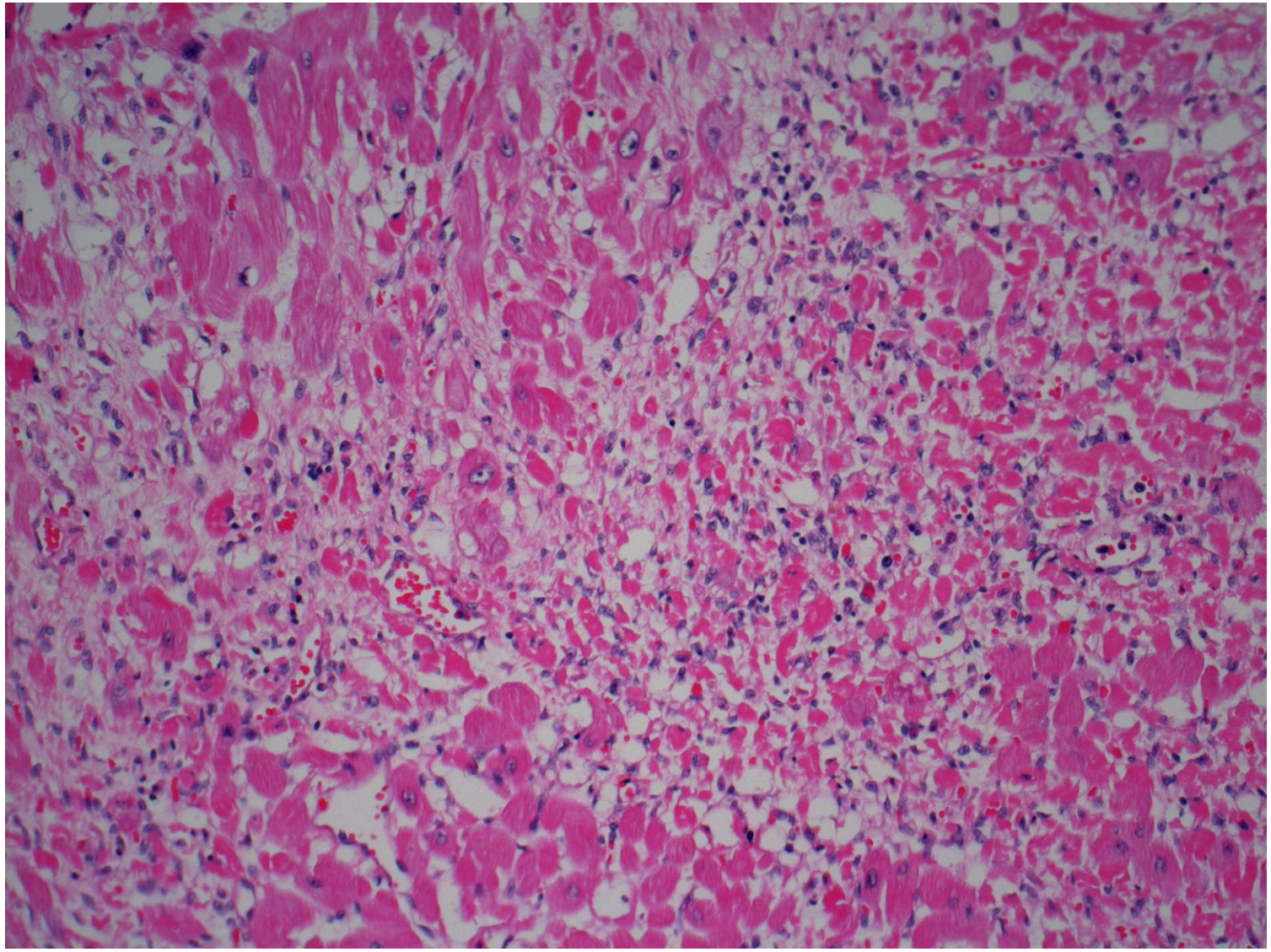
EM features of nemaline body myopathy (16-year-old boy). Electron-dense, lattice-like inclusion bodies are located beneath the subsarcolemmal region. Note the direct continuity of the inclusion bodies to the Z-band. EM-2



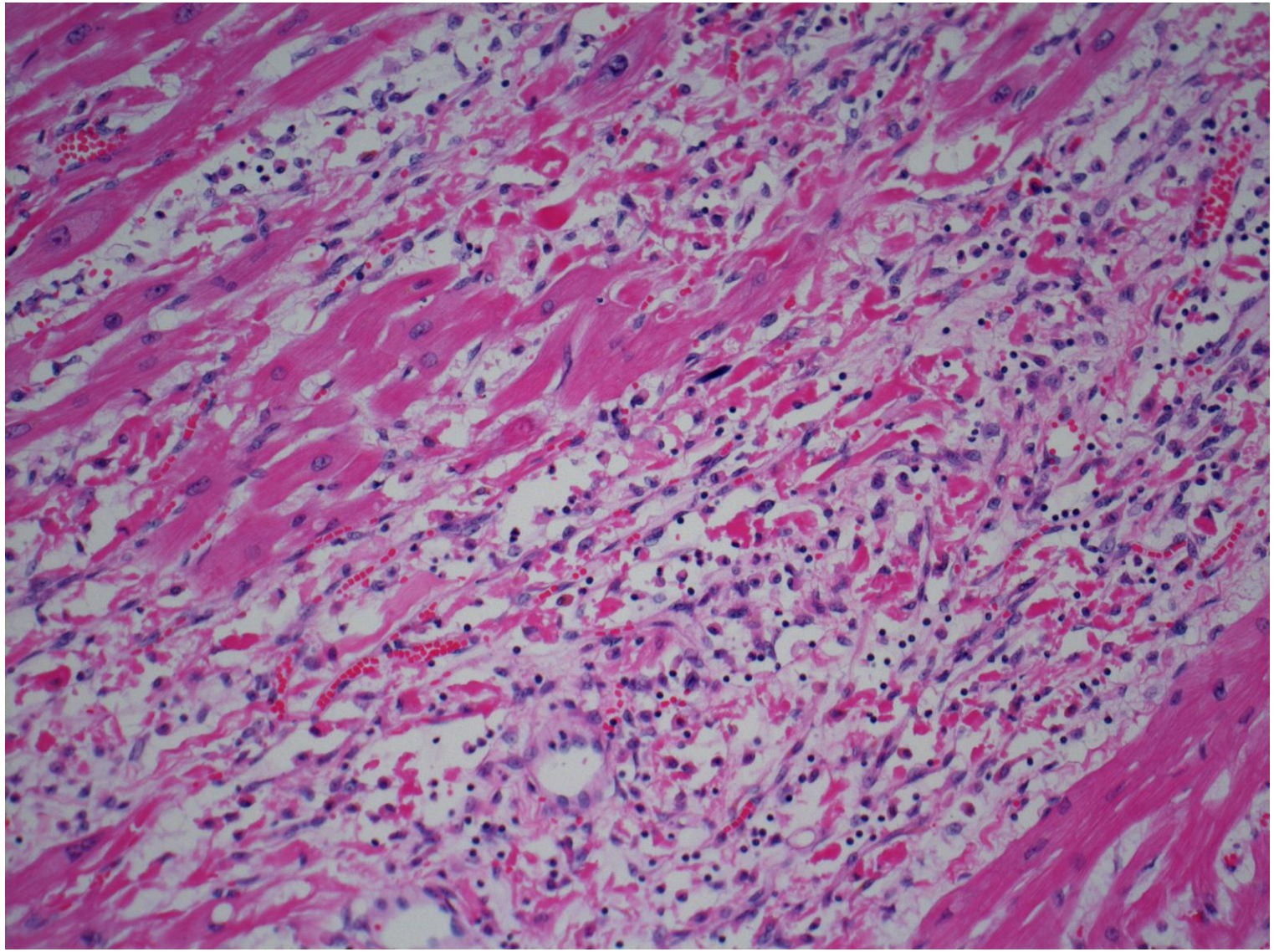
EM features of nemaline body myopathy (16-year-old boy). Electron-dense, lattice-like inclusion bodies are located beneath the subsarcolemmal region. The deposits consist of irregular-shaped and fused collections. EM-3



EM features of nemaline body myopathy (16-year-old boy). Electron-dense, lattice-like inclusion bodies are located beneath the subsarcolemmal region. The lattice structures resemble those of the Z-band. EM-4



Myocarditis was a direct cause of death of this young guy with nemaline myopathy. Incidental lethal viral myocarditis is highly likely. H&E-a



Myocarditis was a direct cause of death of this young guy with nemaline myopathy. Incidental lethal viral myocarditis is highly likely. H&E-b