

Rubinstein Tayby

46 year old man with Rubinstein Tayby syndrome

- Other PMH:
 - Epilepsy
 - Asthma (on home nebuliser)
 - Sleep apnoea (on CPAP)
 - Previous pneumothorax
 - Previous spinal stabilisation (kyphoscoliosis)

46 year old man with Rubinstein Tayby syndrome

- Had not been using CPAP recently because he couldn't tolerate
- Found unresponsive during ward round
- No resuscitation attempts (DNAR)

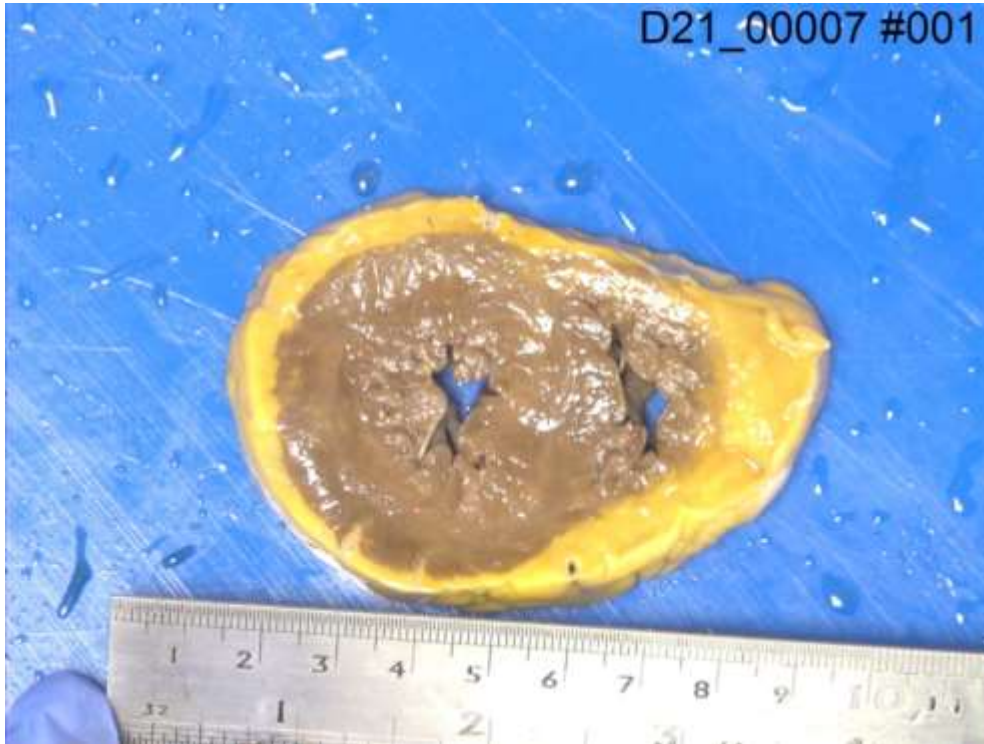
46 year old man with Rubinstein Tayby syndrome

- Neuropath report: No immediate cause of death found in samples examined
- Only features seen consistent with chronic epilepsy

PM findings

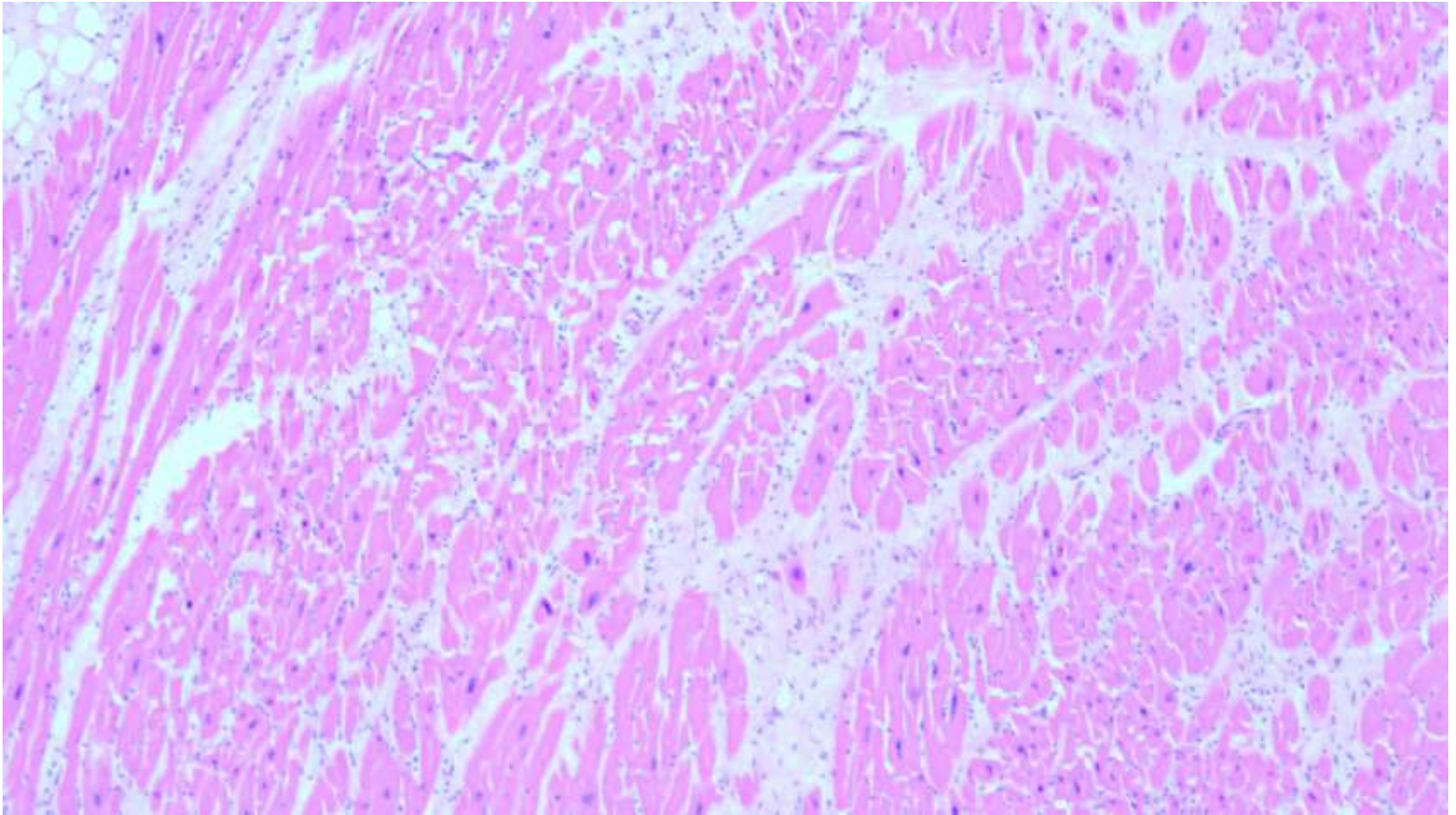
- Pristine coronary arteries
- Mild LVH
- Patent foramen ovale
- Pulmonary congestion, oedema, emphysema
- Left pleural adhesions
- Very long fingers and feet
- Severe scoliosis
- Tiny atrophic kidney right
- Bladder stones

PM heart

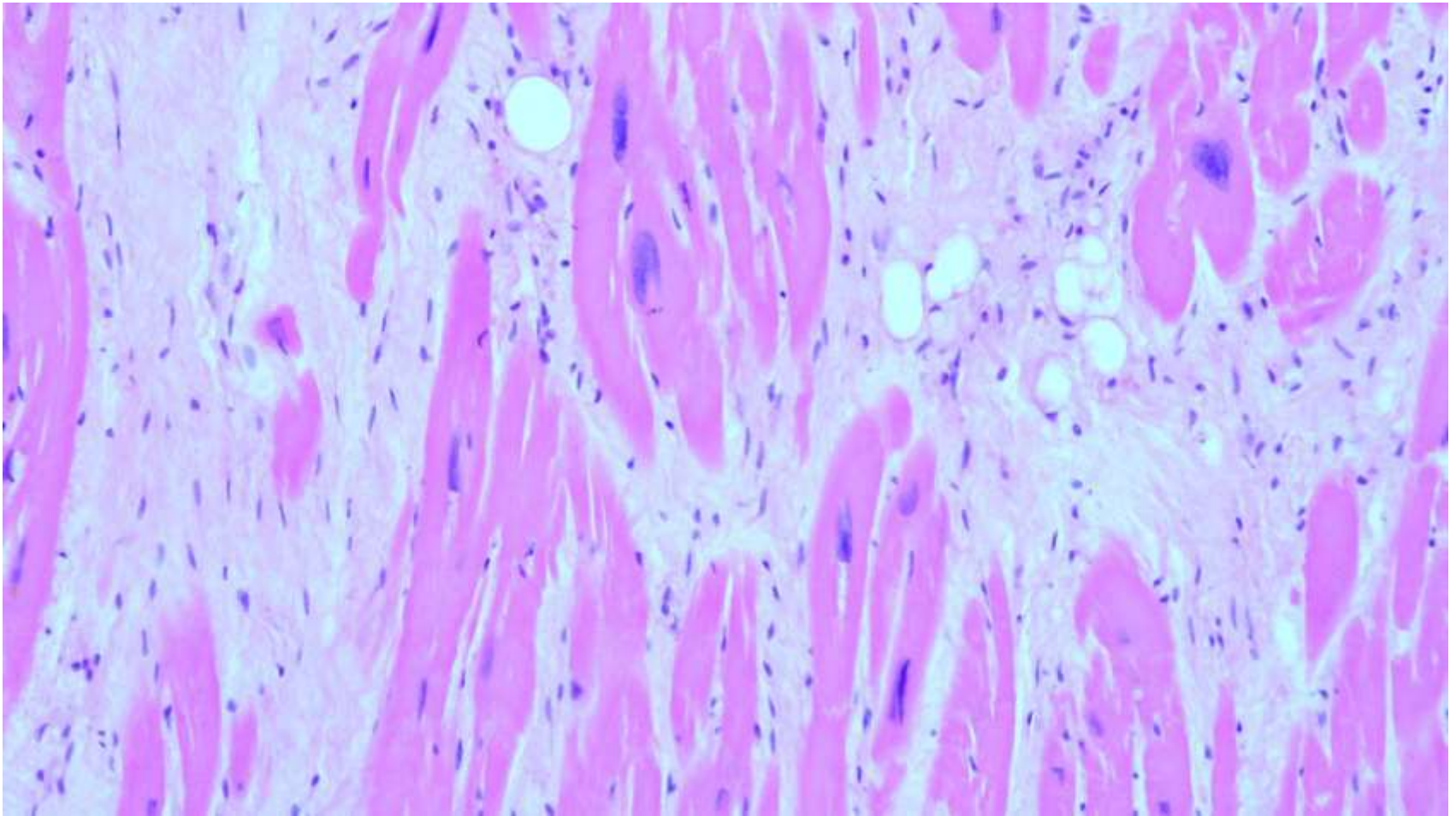


- Mild LVH
- BMI 16.4
- Body weight 42 kg
- Heart weight 334 g

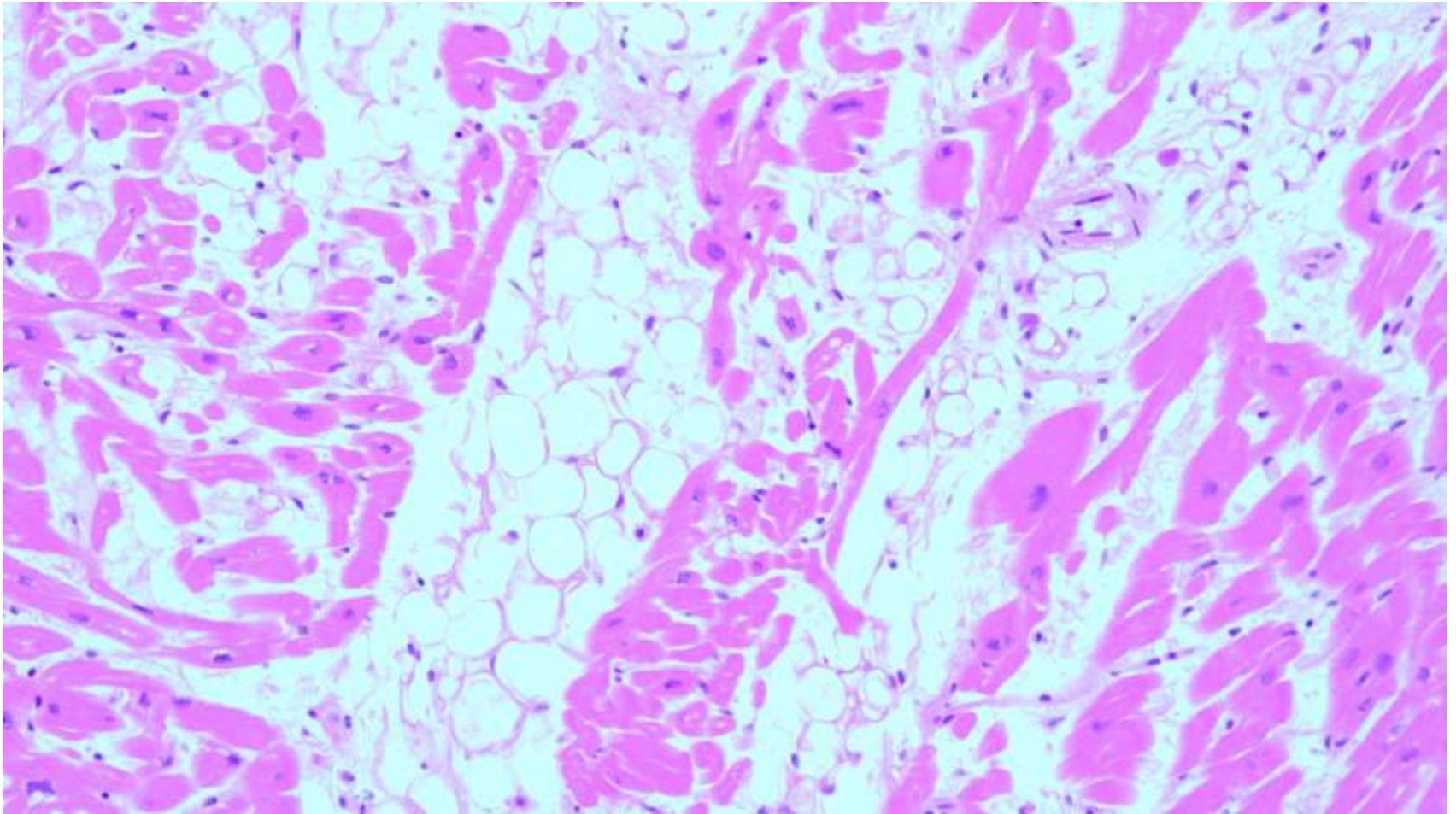
PM heart (focal interstitial fibrosis)



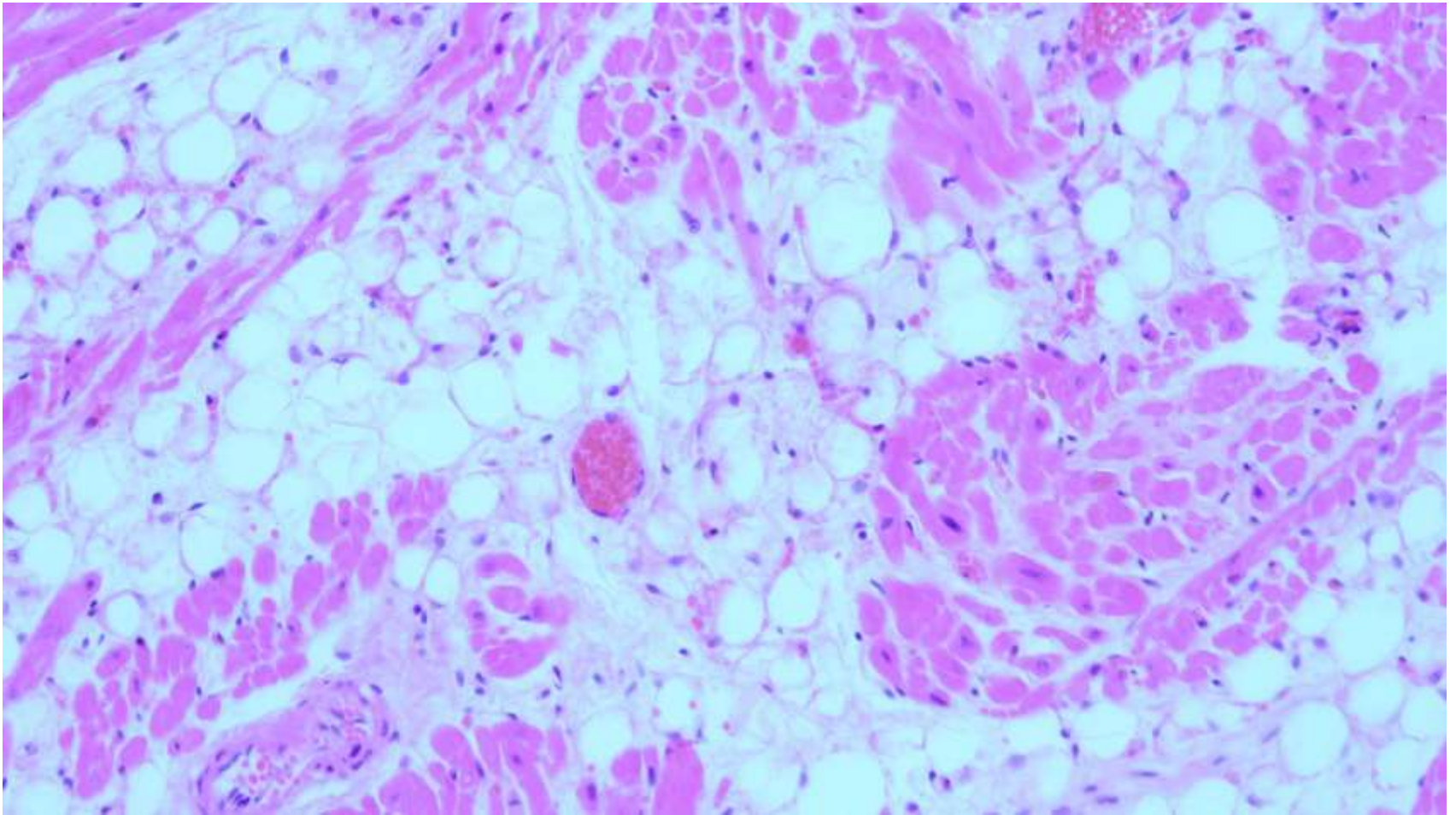
Focal interstitial fibrosis with enlarged cardiomyocyte nuclei



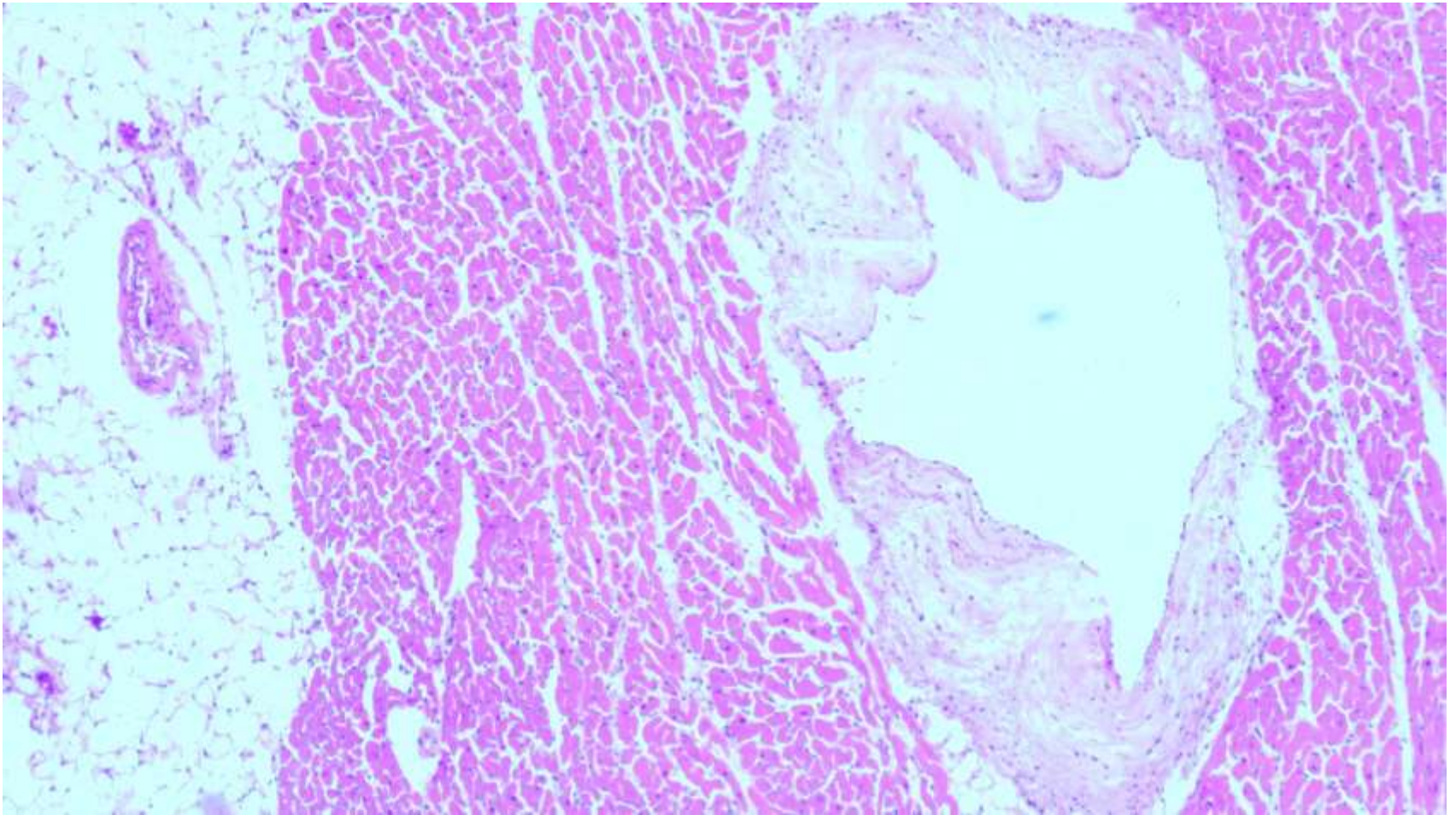
Interstitial fat



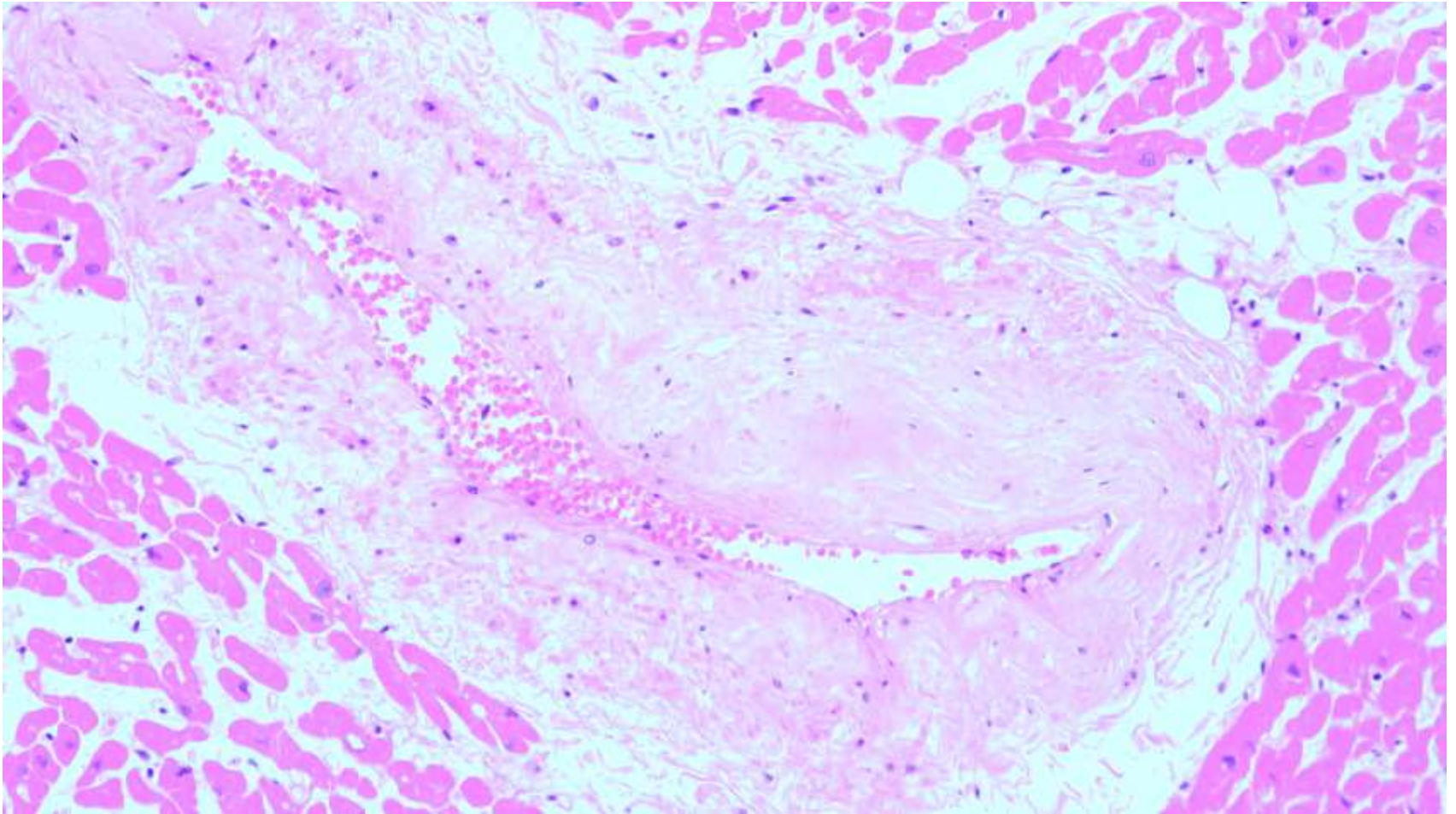
Interstitial fat (? Atrophy)



? Small vessel disease



? Small vessel disease



Questions

- Are the cardiac features sufficient to explain a cardiac death?
- If not, in view of history of sleep apnoea and epilepsy is it reasonable to give a cause of death as just Rubinstein Tayby syndrome without specifying the terminal event further?