Mitral Valve Neurofibroma in A Patient with Neurofibromatosis Type 1: A Rare

A 53-year-old female with neurofibromatosis type 1 (NF-1) was evaluated because of shortness of breath. Her physical examination revealed numerous cafe-au-lait macules (Figure 1A), cutaneous neurofibromas over her whole body (Figure 1B) and axillary freckles (Figure 1C). During her cardiac examination, a grade 3/6 systolic murmur was detected on mitral focus. Echocardiography revealed moderate mitral regurgitation. To conduct a detailed examination, transoesophageal echocardiography was performed. It showed moderate mitral regurgitation (Figure 2) and a 6 × 8 mm anterior mitral valve nodule (Figure 1D). Taking into account the patient’s cutaneous neurofibromas, we assumed that the nodule on the mitral valve was a neurofibroma. Further examination with CT or MRI was suggested, but the patient refused it.

Figure 1. Numerous cafe-au-lait macules (A), cutaneous neurofibromas (B), and axillary freckles (C), transoesophageal echocardiography demonstrated an anterior mitral neurofibroma (D).
Figure 2. Transoesophageal echocardiography demonstrated mitral regurgitation.