Periorbital ecchymosis and shoulder pad sign in transthyretin amyloidosis

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DESCRIPTION

A 75-year-old man was referred for investigation of the aetiology of heart failure. He was admitted for heart failure 5 months prior, and echocardiography showed thickening of the ventricular walls and impaired diastolic function. He had also experienced shoulder stiffness bilaterally for 1 year. Physical examination revealed periorbital puffiness, ecchymosis (figure 1A) and enlarged swelling of the shoulders (shoulder pad sign, figure 1B).

Periorbital ecchymosis and the shoulder pad sign are classically considered pathognomonic of acquired monoclonal immunoglobulin light-chain (AL) amyloidosis. The blood and urinary immunoelectrophoresis and a nephelometric assay of the light chain in serum were examined, but they were negative. Thus, a myocardial biopsy was performed. Amyloid deposits were detected based on positive results of immunofluorescence using transthyretin (TTR) antibodies (figure 2A–D). Molecular analysis of the *TTR* gene was performed, but it revealed no mutations, resulting in a diagnosis of wild-type TTR amyloidosis. The patient has been under consideration for taking tafamidis, an oral TTR tetramer

Figure 2 (A) HE stain. (B) Congo red-stained biopsy of myocardium showing amyloid deposits. (C) Direct fast scarlet stain revealing amyloid deposits. (D) Positive staining for antitranssthyretin antiserum on immunohistochemistry.

stabiliser, which binds to TTR and prevents tetramer dissociation and amyloidogenesis.

Periorbital ecchymosis, also known as raccoon eyes, is observed in amyloidosis. Amyloid deposition leads to vascular fragility and bleeding diathesis is caused by vessel wall damage.¹ In addition, the shoulder pad sign is a typical feature of amyloidosis, which results from amyloid deposition in the periarticular soft tissue of the shoulders. Periarticular swelling is associated with pain, stiffness and decreased range of motion.²

Periorbital ecchymosis and the shoulder pad sign are rarely reported in TTR amyloidosis compared with that in AL amyloidosis.³ However, regardless of the type of amyloidosis, these signs are considered manifestations that may offer diagnostic clues of amyloidosis.

Learning points

- ▶ Periorbital ecchymosis and the shoulder pad sign are typical physical features of amyloidosis.
- ▶ Periorbital ecchymosis and the shoulder pad sign are observed in not only immunoglobulin light-chain amyloidosis but also transthyretin amyloidosis.

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Figure 1 (A) Physical examination showing periorbital puffiness and ecchymosis. (B) Enlarged swelling of the shoulders indicating the shoulder pad sign.

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