Ital. J. Neurol. Sci. 2: 143-148, 1982

Association of cardiomyopathy with Kugelberg-Welander disease.

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A case of Kugelberg-Welander disease with echocardiographic evidence of mitral and tricuspid valve prolapse, in the contest of a cardiomyopathy that might be pathogenically related to the neurologic disease is reported

Key-words: Kugelberg - Welander disease -- cardiomyopathy echocardiography.

Cases of Kugelberg-Welander disease (K-W) with coexisting supraventricular arrhythmias and conduction disturbances, especially atrioventricular block of varying degree [2, 7, 8] are on record. The only report known to us of echocardiographic changes in a K-W patient is of enlargement of the right ventricle and left atrium [9]. We therefore feel that a case of K-W of Dyck HMN type I [1] in which echocardiography yielded findings consistent with cardiomyopathy is worth reporting.

Case report

This 49 year old male with no family history of hereditary disease began as a youngster to feel some loss of power in the lower limbs. He was rejected for military service because of a lower limb motor deficit. At the age of 30 muscular dystrophy was diagnosed. When he was 40 the patient noted a loss of strength in the upper limbs at proximal level accompained by disabling muscular atrophy. By the age of 48 he could walk only with support and 6 months before admission he became bedbound. On neurologic examination he was found to have severe atrophy of the shoulder girdle and arm muscles with grade 0-2 paresis but relatively spared strength (3-4) in the distal muscles. Ankylosis of the elbows prevented the patient from stretching his arms more than 130~. There was diffuse atrophy of the lower musculature and severe paresis (0-2) of the muscles of the pelvic girdle. Strength was conserved more in the distal muscles. Ankylosis of the knees severely restricted extension of the legs. The standing position, maintained only with bilateral support, revealed very marked lumbar lordosis. The deep reflexes were absent in the arms; the knee jerks were absent but ankle jerks were present. The strength of the neck flexor muscles was grade 2 whilst the extensors were normal. The respiratory musculature was normal but the abdominal musculature was severely weakened. There were no pyramidal or cerebellar signs and no cranial nerve deficits. Sensation was intact and the cognitive functions were perfectly normal. The laboratory tests, in particular the inflammatory and metabolic indices and muscle enzymes were normal, as were X-rays of the skull and spine and the EEG. CT showed atrophy of only a few small cortical sulci in the parietal region. The CSF protein pattern, studied with nephelometry and isoelectric focusing, revealed nothing abnormal. The visual and somatosensory evoked potential at wrist and ankle was normal. An electromyographic study was done of the 1st interosseous, gemellus and anterior tibial muscles bilaterally. Every muscle presented fibrillation at rest with abundant polyphasic MUP. There was a voluntary recruiting deficit with evidence of scant 9-10 mV activity. Conduction velocity was normal in the ulnar and common peroneal nerves. Orthodromic sensory conduction velocity, recorded from the right sural nerve, was normal. The left quadriceps muscle was biopsied. For technical reasons it was possible to study only the frozen fragment. Histologic examination revealed a pattern of small muscular fasciculi, made up of generally normotrophic fibers and occasional hypertrophied fi-

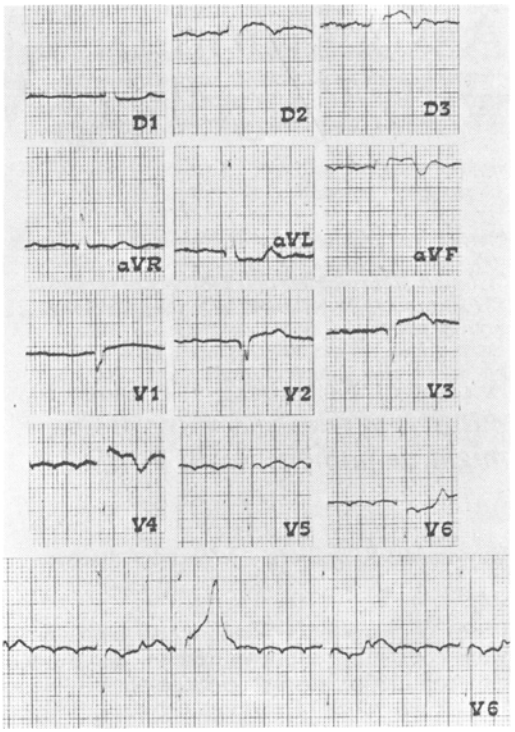

Fig. 1. *12 leads" of* ECG; *see text for explanalion.*

bers with internally located nuclei. The enzyme mosaic was characterised by the presence of both fiber types in homotypic groups. Clinical examination showed marked edema of the face, especially in the periorbital region, turgor of the jugular veins and positive venous pulse. On auscultation of the heart the patient was found to have a grade 4/6 pansystolic regurgitant murmur, audible in the left parasternal region and over the apex, irradiating to the axilla. The heart rate was 48/min and blood pressure 140/90 mmHg. The chest X-ray showed overall enlargement of the heart shadow and congestion of the hili of the lungs. ECG showed atrial flutter with total AV block, high idioventricular rhythm with isolated ectopic beats, extreme left axis deviation as if due to left anterior hemiblock, Q waves in v1, v2 and V3 (Fig. 1). The patient underwent echocardiography with a Smith-Kline Ekoline 20 echocardiograph equipped with transducer focused at 10 cm emitting 2.25 MHz ultrasound waves and with a Smith-Kline Ekoline 21 fiberoptic recorder. The investigation showed pansystolic mitral valve prolapse with multiple echoes in systole and wide excursion of the leaflets in diastole (Fig. 2). The findings in the tricuspid valve (Fig. 3) were similar. The interventricular septum and the posterior wall of the ventricl6 were sluggish with significant impairment of ventricular function (shortening fraction 19%, NV > 29%) (Fig. 4); the dimensions of both ventricles and of the left atrium were at the upper limit of normal.

Discussion

On the clinical, EMG and histopathologic evidence this was a case of Dyck HMN type I or K-W disease. This diagnostic hypothesis is supported by the clinical course with onset in youth and slow but steady deterioration, by the severe atrophy of the shoulder girdle, the absence of sensory disturbances, the frankly neurogenic EMG pattern without slowing of motor or sensory conduction velocity, the normality of the somatosensory evoked potential, the normality of the plasma CPK levels and the histoenzymologic study of the biopsy fragment. Myopathic features in skeletal muscle (hypertrophied fibers with internal nuclei) is not unduly rare in cases of K-W [3]. The absence of a family history is not surprising since one-third of K-W cases are sporadic [1]. In our patient the cardiac chambers were borderline large and yet left ventricular function was impaired. The prolapse of the mitral and tricuspid valve leaflets has a multiple pathogenesis, since it may be due to chordae

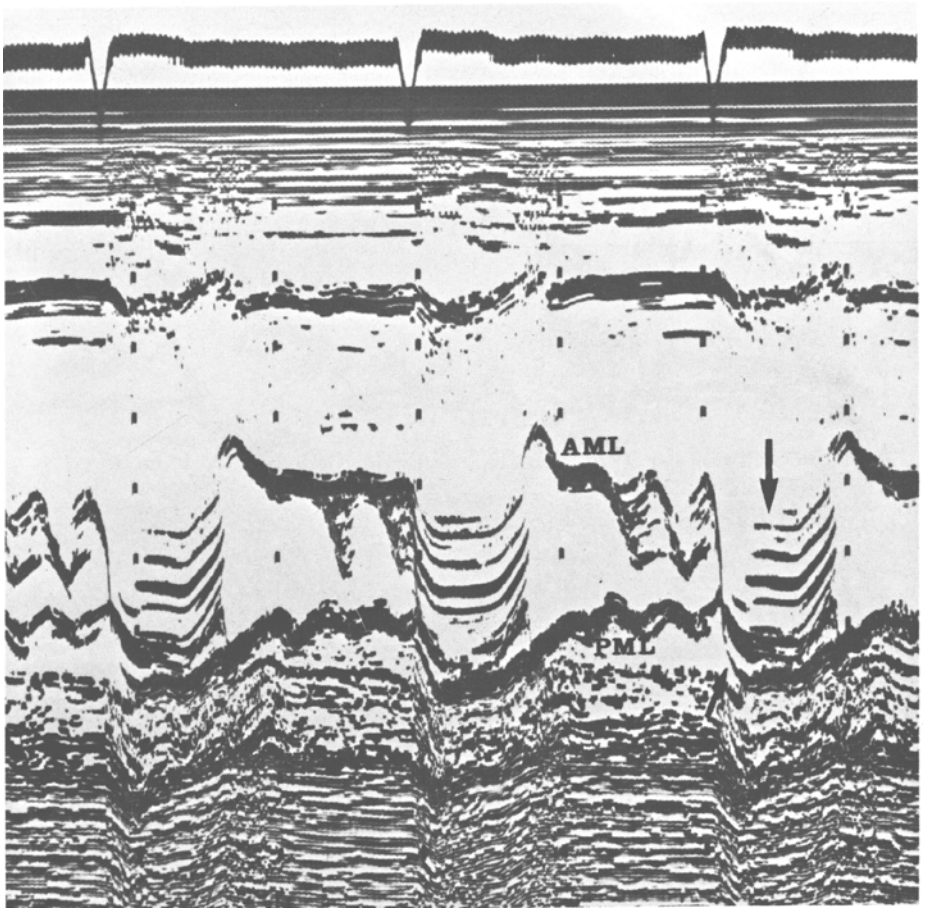

Fig. 2. *Echogram showing pansystolic prolapse (arrow) of both anterior* (AML) *and posterior* (PML) *leaflet of the mitral valve.*

tendineae pathology, to an excess of valve leaflets, to dysfunction of the papillary muscles or of the portion of the myocardium on which they are inserted. In our patient the striking multiple echoes in systole could be compatible with valve leaflet redundancy secondary to myxomatous degeneration of same [4, 6]. On the other hand, ultrasound findings like ours reflect degenerative processess in the papillary muscles, in the presence of normal atrioventricular valves [5]. Tanaka at al [9, 10] found interstitial fibrosis of the walls of the right ventricle and ultrastructural changes in the myocardial fibers, chiefly loss of myosin filaments, in patients with K-W. Further, the impairment of ventricular function and the ECG changes found in our patient are compatible with the existence of a cardiomyopathy for which there is nothing in the history to indicate another etiology. Indeed the patient had previously presented no clinical signs or symptoms of cardiopathy, was normotensive, had no collagen disease and had nothing in his history that might point a toxic or infective heart disease. In view of these findings we consider that the valvular dysfunction found in our patient may have been the outcome of primary disease of the papillary muscles or of the area of the ventricles on which they are inserted. However, only systematic investigation of cardiac function in patients with K-W disease can disclose the percentage of cases with cardiac alterations and establish whether they have a common pathogenesis or whether they are simply coincidental.

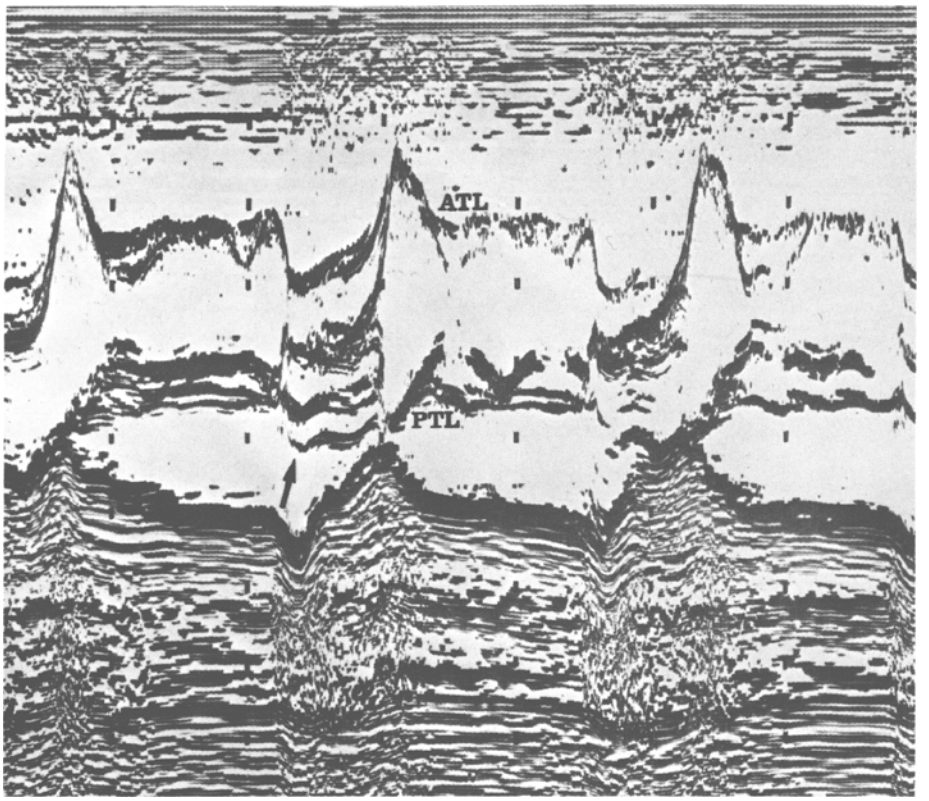

Fig. 3.

Fig. 3. *Echogram showing pansystolic prolapse (arrow) of posterior tricuspid leaflet* (PTL), *with exaggerated motion of anterior tricuspid leaflet* (ATL) *in diastole.*

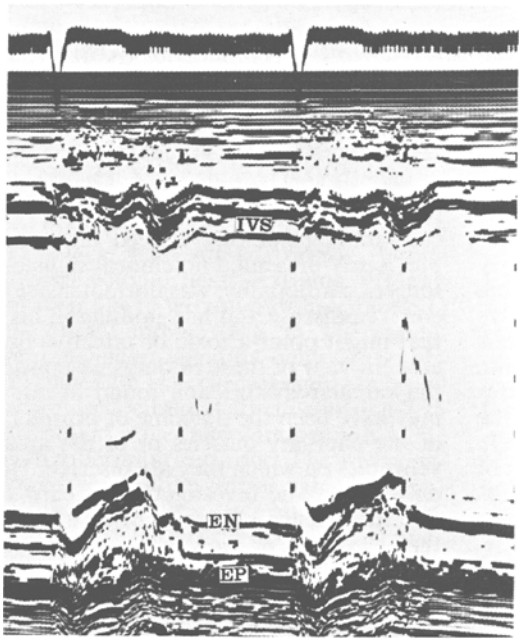

Fig. 4.

Fig. 4. *Echogram of the left ventricle shoving reduced movement of both interventricular septum* (IVS) *and posterior endocardium* (EN) *and epicardium* (EP). *The finding is consistent with impaired contractility.*

Sommario

Viene presentato un caso di malattia di K-W reperti ecocardiografici di prolasso delle valvole mitrale e tricuspide nell'ambito di una cardiomiopatia che potrebbe essere patogeneticamente correlata alia malattia neurologica.

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