A Case of the Kugelberg-Welander Syndrome Complicated with Cardiac Lesions

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SUMMARY

There have been a few reports of cardiac involvement in the Kugelberg-Welander syndrome. We presented a case of this syndrome complicated with cardiomegaly and conduction disturbance. The patient, a 21-year-old woman, had atrial standstill and A-V junctional rhythm. The chest X-ray film showed marked cardiomegaly. The His bundle electrogram revealed that H-V interval was 40 msec. She was treated with implantation of cardiac pacemaker.

Additional Indexing Words:

Muscle atrophy Cardiomegaly Atrial standstill Junctional escape rhythm

THE Kugelberg-Welander syndrome was described by Wohlfart et al,[1] and Kugelberg and Welander[2] as a heredofamilial juvenile muscular atrophy simulating muscular dystrophy. The characteristic features of this syndrome are the followings; onset in childhood or adolescence; atrophy and weakness of mainly proximal limb muscles; slowly progressive clinical course; development of fasciculation; evidence of neurogenic changes in electromyogram and muscle biopsy; nonsex-linked recessive inheritance. Since then, there have been many reports on the Kugelberg-Welander syndrome. In some of them, fasciculation was not seen, and variable modes of inheritance were noted. Sporadic occurrence was also noted.

It is well known that cardiac involvements are frequently found in neuromuscular diseases, such as Duchenne's muscular dystrophy,[3] myotonic dystrophy,[4] Friedreich's ataxia,[5] and so on. However, there have been only a few reports on the Kugelberg-Welander syndrome with cardiac lesions. In this paper we presented a case of the Kugelberg-Welander syndrome with cardiac enlargement and abnormalities in the electrocardiogram.

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Received for publication September 21, 1979.

Case Report

A 21-year-old woman was admitted to this hospital, because of muscle atrophy and weakness in October, 1978. She had apparently been well until 4 years of age, when it was noted that she walked awkwardly and she could not run fast. She was diagnosed to have progressive neuromuscular dystrophy when she was 9 years old. However, she had not been restricted in her every-day life. Three months before admission she was found to have bradycardia, and the chest X-ray film at that time showed cardiac enlargement. There was no evidence of neuromuscular diseases in her family.

Physical examination on admission revealed a slender woman. The height was 155 cm. The body weight was 40 Kg. The pulse was 40/min and regular. The blood pressure was 136/60 mmHg. A grade 2/6 systolic ejection murmur was heard at the apex. Liver and spleen were not palpable. Neurological examination disclosed moderate atrophy and weakness in the proximal portion of the arms and legs. Mental state and intelligence were normal. There was no abnormality in the cranial nerves. Scapula arata and pseudohypertrophy were not seen. Fasciculation was not observed. Achilles tendon and knee jerk reflexes were lost. Abdominal reflexes were normal. Sensory examination was normal. No abnormal reflexes were seen. The gait was waddling.

The serum electrolytes, glutamic oxaloacetic transaminase, glutamic pyruvic transaminase, lactic dehydrase, protein fraction, cholesterol, β-lipoprotein, triglyceride, and blood sugar were all within normal range. Serum creatine phosphokinase was increased (126 units). Serum creatinine was decreased (0.56 mg/100 ml). Serum creatine and urine creatine were increased (0.58 mg/100 ml, 16.9 mg/100 ml, respectively). Renal function and respiratory function were normal. Cerebrospinal fluid was normal. The chest X-ray film (Fig. 1) on admission revealed marked cardiomegaly compared to the one taken about 1 year previously. The electro-

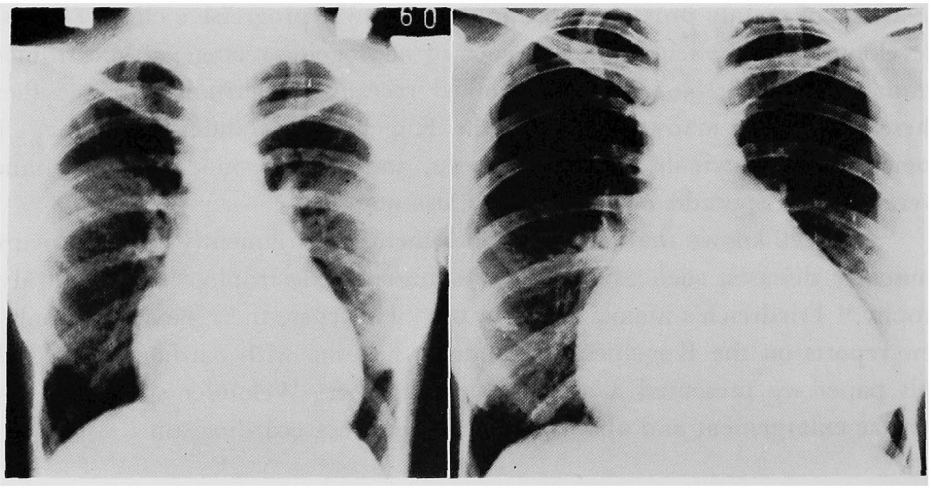

Fig. 1. Chest X-ray film on admission (right, October, 1978) showing marked cardiomegaly compared to the one taken about 1 year previously (left, July, 1977).

cardiogram (Fig. 2) showed atrial standstill and A-V junctional rhythm. Left axis deviation and left ventricular hypertrophy were noted. The echocardiogram disclosed no abnormalities of mitral valve, and thickness of septum and posterior wall was within normal range. Left ventricular diastolic dimension was slightly in-

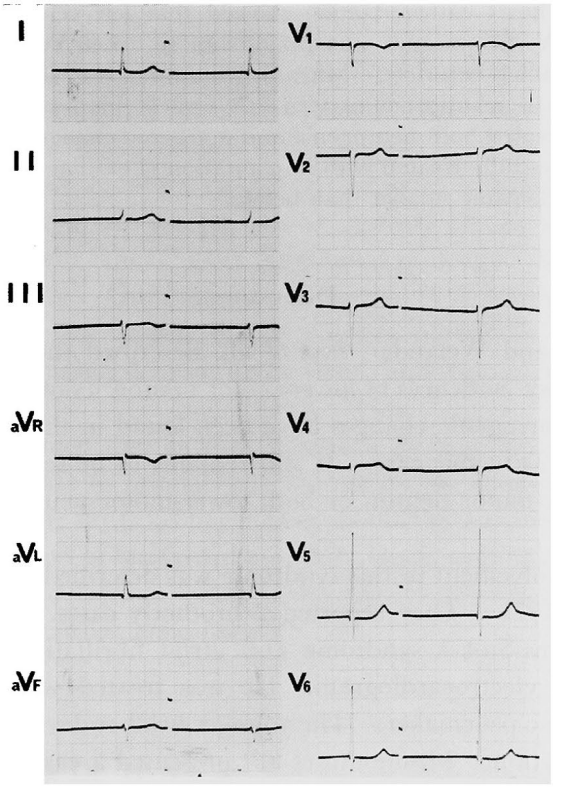

Fig. 2. Electrocardiogram on admission showing atrial standstill, A-V junctional rhythm, left axis deviation and left ventricular hypertrophy. Heart rate is 40/min.

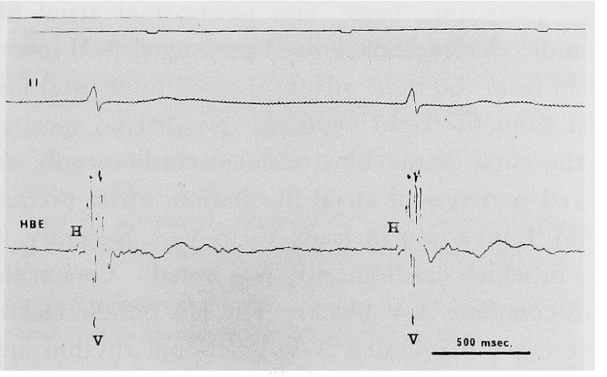

Fig. 3. His bundle electrogram and electrocardiogram (lead II). H-V time is within normal range (40 msec). A wave is not seen.

creased (57 mm). Ejection fraction was 0.77. The His bundle electrogram (Fig. 3) showed a His bundle potential (H) preceding each V wave. A wave was not seen. The H-V interval was 40 msec. Right heart catheterization revealed that pulmonary capillary wedge pressure was 16 mmHg, pulmonary artery pressure 34/16 mmHg, right ventricular pressure 46/16 mmHg, mean right atrial pressure 13 mmHg. Left heart catheterization showed that left ventricular pressure was 152/18 mmHg. Aortic pressure was 152/68 mmHg. Left ventriculogram showed good contraction of left ventricle. Mitral regurgitation was not seen. The electromyogram showed a neurogenic pattern. A muscle biopsy specimen from right deltoid muscle revealed both neurogenic and myogenic changes.

She was successfully treated with implantation of cardiac pacemaker, by which cardiac silhouette became smaller than before.

Discussion

Kugelberg and Welander, first of all, described the electromyographic and muscle biopsy evidences of lower-motor neuron lesion.[2] However, both neurogenic and myogenic changes have been found in the skeletal muscles in this syndrome,[6] as in our case. It still remains to be seen whether primary lesion is in lower-motor neuron or both lower-motor neuron and the skeletal muscle.

Cardiac involvement in this syndrome was described for the first time by Sterz et al[7] in 1971. They reported 2 brotherly cases. The older brother manifested Adams-Stokes syndrome and atrial fibrillation with A-V block was seen in his electrocardiogram. He was treated with implantation of permanent cardiac pacemaker. The younger brother developed cardiomegaly and atrial fibrillation. Masumoto et al[8] presented a case in which the chest X-ray film showed cardiomegaly and the electrocardiogram revealed atrial standstill, A-V junctional rhythm and left ventricular hypertrophy, as seen in our case. Sugimura et al[9] reported 2 cases. In one of them, first degree A-V block, atrial premature beats, and paroxysmal atrial fibrillation were seen, and His bundle electrogram showed prolonged A-H interval. A biopsy specimen obtained from the right atrium showed interstitial fibrosis, although the one obtained from the right ventricle revealed no specific changes. In the other case, the chest X-ray film revealed cardiomegaly and the electrocardiogram showed paroxysmal atrial fibrillation, atrial premature beats, left axis deviation and T inversion in leads V_4 to V_6. Recently, Tanaka et al[10] described 2 cases in which cardiomegaly was noted. One of them manifested atrial flutter with complete A-V block. The His bundle electrogram showed A-H block. The other manifested A-V junctional rhythm and abnormal Q wave in leads I, aVF, and V_5 to V_6. The His bundle electrogram showed normal. A biopsy specimen from the right ventricle revealed a slight inter-

stitial fibrosis.

Among 8 cases, abnormalities in the electrocardiogram were seen in all cases, and cardiomegaly was noted in 6 cases of them. Furthermore, endocardial biopsy specimen from the right atrium and the right ventricle (Sugimura's case[9] and Tanaka's case[10]) showed the pathologic changes. These facts suggest that organic lesions were present in atrial and ventricular myocardium in this syndrome. In addition, the fact that the atrial arrhythmias and A-V block are frequently found in this syndrome may indicate that the myocardial changes strongly influence the atrium and the conducting system.[9),10] Furthermore, among 4 cases reported so far including ours in which the His bundle electrogram was performed, normal His bundle electrogram was noted in 1 case, the other 2 cases manifested prolonged A-H interval and A-H block, respectively. Our case showed normal H-V interval, and A wave was not seen. This fact suggest that the upper portion in the conducting system is apt to be damaged more easily than the lower portion in this syndrome.

Since we confirmed that cardiac silhouette became smaller than before by increasing heart rate to normal with temporary pacemaker, we think that the implantation of the artificial pacemaker should be considered in the case who has a complication of bradycardia or A-V block, even if the myocardium might be also damaged, because bradycardia might further deteriorate cardiac function and induce cardiac enlargement.

Acknowledgments

The authors thank Dr. H. Yasuda and Dr. H. Nishijima of Hokkaido University for their help.

References

1. Wohlfart G, Fex J, Eliasson S: Hereditary proximal spinal muscular atrophy: a clinical entity simulating progressive muscular dystrophy. Acta Psychiat Neurol Scand **30**: 395, 1955
2. Kugelberg S, Welander L: Heredofamilial juvenile muscular atrophy simulating muscular dystrophy. Arch Neurol Psychiat **75**: 500, 1956
3. Perloff JK, De Leon AC, O'Doherty D: The cardiomyopathy of progressive muscular dystrophy. Circulation **33**: 625, 1966
4. Uemura N, Tanaka H, Niimura T, Hashiguchi N, Yoshimura M, Terashi S, Kanehisa T: Electrophysiological and histological abnormalities of the heart in myotonic dystrophy. Am Heart J **86**: 616, 1973
5. Thoren C: Cardiomyopathy in Friedreich's ataxia. With studies of cardiovascular and respiratory function. Acta Paediat Scand **153** (suppl): 1, 1964
6. Hausmanowa-Petrusewicz I, Askanas W, Badurska B, Emeryk B, Fiazianska A, Garbalinska W, Hetnarska L, Jéarzgjowska H, Kamieniecka Z, Niebroj-Dobosz I, Prot J, Sawicka E: Infantile and juvenile spinal muscular atrophy. J Neurol Sci **6**: 269, 1968

7. Sterz H, Harrer G, Marchet H, Kaserer HP, Schlamberger H, Samec HJ, Stark U: Premäre und neurogene Skeletmuskelerkrankungen bzw.-paralysen mit schweren kardialen Rhythumstörungen. Z Kreislaufforsch **60**: 1, 1971
8. Matsumoto K, Kakiuchi F, Kakihana M, Katayama S, Sakamoto T, Murao S: Kugelberg-Welander disease with cardiopathy of unknown etiology. Clinical report of a case. Respiration and Circulation **19**: 863, 1971 (in Japanese with English abstract)
9. Sugimura F, Iijima M, Ozawa Y, Ooki Y, Watanabe S: Two cases of Kugelberg-Welander disease with cardiopathy. Clin Neurol (Tokyo) **13**: 79, 1973 (in Japanese with English abstr)
10. Tanaka H, Uemura N, Toyama Y, Kudo A, Ohkatsu Y, Kanehisa T: Cardiac involvement in the Kugelberg-Welander syndrome. Am J Cardiol **38**: 528, 1976