Familial secundum atrial septal defect with dysrhythmia associated with web neck

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Most cases of atrial septal defect occur sporadically, but a few families have the defect as a genetic abnormality. A family having familial type secundum atrial septal defect with dysrhythmia associated with web neck is reported. In this family, two female siblings aged 11 (Case 1) and 4 years (Case 2) and their father had secundum atrial septal defect. Case 1 presented with two year history of syncope attacks and Case 2 with easy fatigability since early childhood. Both sisters also had web neck as a solitary anomaly. Electrocardiograms revealed prolonged PR interval and right bundle-branch block in both cases. In Case 1 first-degree atrioventricular block and Mobitz type I and II block were observed in Holter monitoring. Echocardiographical examination showed secundum atrial septal defect in both sisters. A permanent pacemaker was implanted in Case 1, and then atrial septal defects in both patients were surgically repaired; no postoperative complaints were observed. The father had been diagnosed as having atrial septal defect when he was 35 years sold, and first-degree atrioventricular block and atrial flutter developed after open heart surgery. In conclusion, the association of secundum atrial septal defect and prolongation of PR interval should be considered as familial occurrence of atrial septal defect. Identification of atrial septal defect in more than one family member should prompt clinical evaluation of all relatives.

Key words: dysrhythmia, familial atrial septal defect, web neck.

Secundum atrial septal defect (ASD) is one of the more common congenital cardiac defects to occur as an isolated lesion. It represents about 6 to 10% of all cardiac anomalies encountered and is more frequent in females than in males (2:1). It is estimated that an ASD occurs in 1:1,500 live births[1]. Most cases of ASD occur sporadically; however, a few families have the defects as a genetic abnormality. Autosomal dominant inheritance is possible for ASD with or without atrioventricular block[2,3]. Familial forms are characterized by the same type of ASD, and are frequently associated with other cardiac, osteoarthricular (Holt-Oram syndrome) or atrioventricular conduction abnormalities[4].

The electrocardiogram (ECG) in ASD usually reveals normal sinus rhythm; however, in a small number of patients, usually older, atrial dysrhythmias including atrial fibrillation, atrial flutter, conduction abnormalities, and sick sinus syndrome may be observed in the natural history of ASD, as well as after open heart surgery[1,5,6].

Herein, we report a family having familial type secundum ASD with conduction defects and atrial dysrhythmia associated with web neck.

Case Reports

Case 1

An 11-year-old girl was admitted with syncope attacks and palpitation. She had two-year history of palpitation attacks during effort and excitement followed by unconsciousness lasting for aproximately five minutes. She was the first child of a nonconsanguineous 41-year-old mother and 37-year-old father. Her father's past history revealed that he had been diagnosed as having secundum ASD when he was 35 years

old, and his defect had been repaired by open heart surgery. Complete atrioventricular block, atrial fibrillation and atrial flutter had developed one year after the operation, and a permanent pacemaker was implanted. Case 1 had a four-year-old sister with secundum ASD and web neck and a 10-year-old healthy sister.

On physical examination, she was conscious, and her general condition was well, with a heart rate of 96/min and blood pressure of 90/60 mmHg. Web neck deformity and low hair-line were present (Fig. 1). On cardiac auscultation, fixed splitting of the second heart sound in addition to the grade 2/6 systolic ejection murmur at the second left intercostal space was heard.

Chest X-ray showed a prominent pulmonary trunk. ECG showed sinusal bradycardia, first-degree atrioventricular block with a PR interval of 0.28 s, and incomplete right bundle-branch block (Fig. 2). On echocardiographic examination, an ASD of secundum type with a dimension of 12 mm at the region of fossa ovale, paradoxal septal movement, and left-to-right shunt at the atrial level were detected. Sinusal bradycardia (minimum heart rate 45/min), first-degree atrioventricular block and Mobitz type I and type II blocks were observed in Holter monitoring. Ultrasongraphic examinations of the abdomen and pelvis were normal. Chromosome analysis showed normal 46, XX pattern.

Syncope attacks of the patient were controlled with the implantation of a permanent pacemaker, and then her ASD was surgically repaired. Now the patient is well with no recurrence of her complaints.

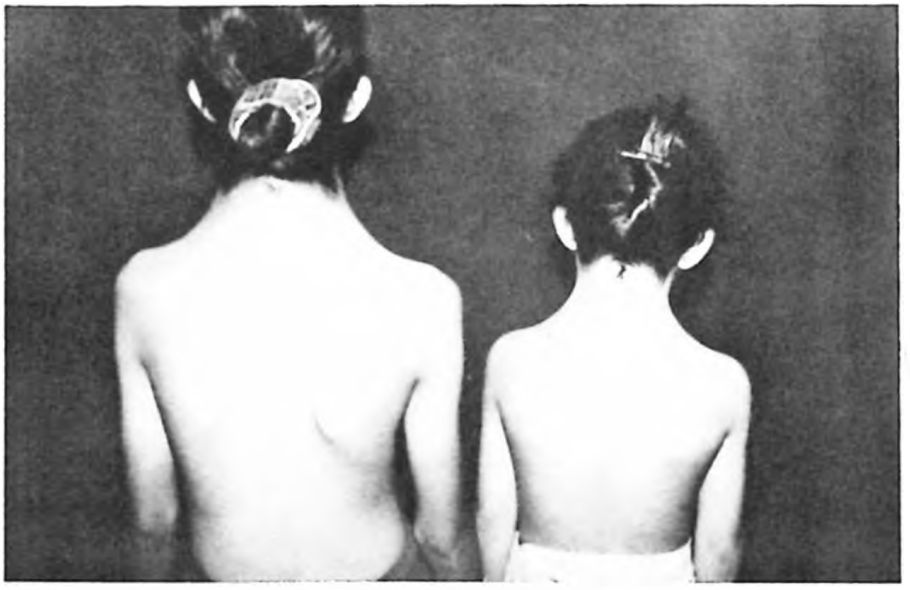

Fig. 1. Web neck deformities and low hair lines of the patients.

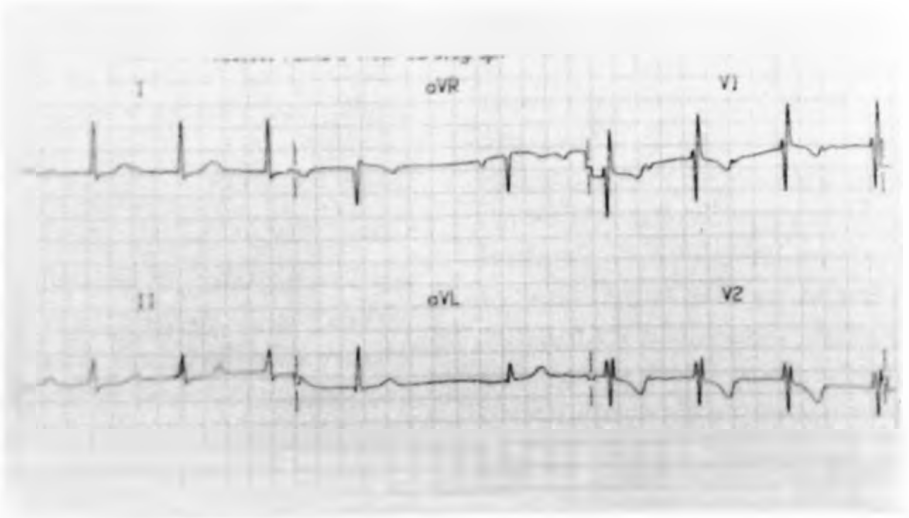

Fig. 2. Electrocardiogram (ECG) of Case 1 showing the prolongation of the PR interval and incomplete right bundle-branch block.

Case 2

A four-year-old female patient and the sister of Case 1 was admitted with easy fatigability recognized since early childhood. On physical examination, she was conscious, and her general condition was well with a heart rate of 92/min and blood pressure of 100/60 mmHg. Web neck deformity and low hair line were present (Fig. 1). On cardiac auscultation, fixed splitting of the second heart sound in addition to the grade 2-3/6 systolic ejection murmur at the second left intercostal space was heard.

Chest X-ray showed increased pulmonary vascular marking. ECG revealed incomplete right bundle-branch block and prolonged PR interval of 0.22 s. On echocardiographic examination, an ASD of the secundum type with a dimension of 13-14 mm at the region of fossa ovale, paradoxal septal movement, dominance of the right atrium and right ventricle, and left-to-right shunt at the atrial level were detected. Chromosome analysis showed normal 46, XX pattern. ASD of the patient was surgically repaired and no postoperative complaints were observed.

Discussion

Although most cases of ASD occur sporadically, the defect may show a familial pattern[1,2]. Autosomal dominant inheritance is possible for these cases[2,3]. The presence of the defect in both the father and his two daughters in the presented family suggests an autosomal dominant trait. Only a few families have been reported in whom the ASD is transmitted according to an autosomal dominant inheritance[2,3,6].

The ECG in ASD usually reveals normal sinus rhythm; however, in a small number of patients, usually older, a junctional rhythm or a supraventricular tachyarrhythmia, such as atrial flutter, can be seen. In most patients, the mean frontal plain QRS axis is to the right, from +90 to +170 °. The PR interval may be prolonged, especially in older patients, because of intraatrial and sometimes H-V conduction delay that results in first-degree atrioventricular block[1]. Prolonged PR intervals have been found in 5-15% of cases of ASD[2]. However, the incidence of PR prolongation in familial cases of ASD has been reported to be between 75-100%[8-11]. According to this literature knowledge, and from our findings, it seems that prolongation of atrioventricular conduction is more common among familial cases of ASD. Thus, the association of secundum ASD and prolongation of PR interval should be considered as familial occurrence of ASD. This point should be considered in genetic counseling. In contrast, some authors have reported some families with secundum ASD without PR prolongation[3,7]. Lynch et al.[12] suggested the existence of at least two distinct hereditary varieties of ASD, one with and one without a prolonged PR interval. Both of our cases and their father had prolonged PR interval in ECG, and one of the siblings (Case 1) also had Mobitz type I and type II blocks. In about half the cases, P wave changes suggest right atrial enlargement. There is also some variant of the rsR' or RSR' pattern ("incomplete right bundle-branch block" pattern) in lead V_1, consistent with right ventricle volume overload, as seen in our two both patients. The duration of the QRS complex is less than or equal to 0.1 second, and R' in lead V_1 is somewhat prolonged[1].

Atrial arrhythmias are the most common late problems following closure of the defect. But unrepaired ASDs are also associated with atrial dysrhythmia, particularly in older patients. Atrial fibrillation and atrial flutter are not common complications of ASD in children but are seen often in adult patients[5]. The father of our patients had experienced complete atrioventricular block, atrial fibrillation and atrial flutter during the postoperative period.

Sinus node dysfunction is one of the complications following surgery for ASD[1,2]. In the past, it has been thought that this arrhythmia was the consequence of either the surgical trauma to the artery supplying blood to the sinus node or damage to the sinus node area[13]. However, the sinus node function of these patients has rarely been investigated preoperatively. Some studies based on a standard ECG were able to detect sinus node dysfunction preoperatively in a limited number of patients[14]. However, preoperative electrophysiologic measurements in patients of all ages detect conduction abnormalities in as many as 40% of patients[1,5,15,16]. Apparent sinus node dysfunction has been reported as a result of finding abnormal corrected sinus node recovery times and sinoatrial conduction times; however, clinically, only rare patients have abnormal findings in resting ECGs or on 24-hour ambulatory monitoring. Perhaps these abnormal electro-

physiologic findings are due to an imbalance of the autonomic nervous system control of the sinoatrial and atrioventricular nodes. Intraatrial conduction time is prolonged in the older patient, and right atrial effective refractory periods are increased in some patients. Patients with both findings may be predisposed to atrial arrhythmias[1].

In addition, both of our cases had web neck deformity which might have been a solitary anomaly or associated with a genetic syndrome such as Ullrich-Turner, Noonan or Down syndromes[17]. Since our cases did not have other phenotypic features of these syndromes, was believed that they had web neck deformity as a solitary anomaly. It has been reported that 60% of infants with web neck had congenital heart defect, with a high incidence of flow-related defects such as hypoplastic left heart, coarctation, and secundum ASD[17]. This association implies a pathogenetic relationship and appears to be independent of causal factors[17]. Therefore, the finding of web neck on a prenatal ultrasound or newborn examination should prompt a search for congenital heart defect.

Familial secundum ASD is probably more frequent than commonly reported since cardiological examination of the relatives is not routinely performed in every case of apparently sporadic ASD. In conclusion, identification of prolongation of the PR interval in a case of secundum ASD should prompt clinical evaluation of all relatives. In addition, all patients with ASD should be carefully evaluated for conduction defects and atrial dysrhythmias.

REFERENCES

1. Porter CJ, Feldt RH, Edwards WD, Seward JB, Schaff HV. Atrial septal defects. In: Emmanouilides GC, Riemenschneider TA, Allen HID, Gutgesell HP (eds). Moss and Adams Heart Disease in Infants, Children, and Adolescents (5th ed) Vol. 1. Baltimore: Williams&Wilkins; 1995: 687-703.
2. Günal N, Gül S, Kahramanyol Ö. Familial atrial septal defect with prolonged atrioventricular conduction. Acta Paediatr Jpn 1997; 39: 634-636.
3. Li Volti S, Distefano G, Garozzo R, Romeo MG, Sciacca P, Mollica F. Autosomal dominant atrial septal defect of ostium secundum type. Report of three families. Ann Genet 1991; 34: 14-18.
4. Cachat F, Rapatsalahy A, Sekarski N, Hurni M, von Segesesr L, Payot M. Three different types of atrial septal defects in same family (Abstract). Arch Mal Coeur Vaiss 1999; 92: 667-669.
5. Fyler DC, Atrial septal defect secundum. In: Fyler DC (ed). Nadas' Pediatric Cardiology. Philadelphia: Hanley&Belfus, Inc.; 1992: 513-524.
6. Friedli B. Arrhythmias in adolescent and adult with a congenital heart defect. Schweiz Med Wochenschr 1993; 123: 2065-2071.
7. Tsuchioka Y, Kawagoe T, Hondo T, et al. Secundum atrial septal defect in two families (Abstract). Kokyu To Junkan 1990; 38: 93-96.
8. Bizarro RO, Callahan JA, Feldt RH, Kurland LT, Gordon H, Bradenburg RO. Familial atrial septal defect with prolonged atrioventricular conduction: a syndrome showing the autosomal dominant pattern of inheritance. Circulation 1970; 41: 677-683.
9. Weil MH, Allenstein BJ. A report of congenital heart disease in five members of one family. N Engl J Med 1961; 265: 661-666.
10. Amarasingham R, Heming HA. Congenital heart disease with arrhythmia in a family. Br Heart J 1967; 29: 78-80.
11. Kahler RL, Braunwald E, Plauth WH. Familial congenital heart disease: familial occurrence of atrial septal defect with atrioventricular conduction abnormalities; supravalvular aortic and pulmonic stenosis and ventricular septal defect. Am J Med 1966; 40: 384-390.
12. Lynch HT, Bachenberg K, Harris RE, Becker W. Hereditary atrial septal defect. Update of a large kindred. Am J Dis Child 1978; 132: 600-604.
13. Tung KS, James TN, Effler DB, McCormack LJ. Injury to the sinus node in open heart operations. J Thorac Cardiovasc Surg 1967; 53: 814-829.
14. Siltanen P. Atrial septal defect of secundum type in adults. Clinical and hemodynamic studies of 129 cases before and after surgical correction under cardiopulmonary bypass. Acta Med Scand 1968; 183-184 (Suppl): 497-520.
15. Bolens M, Friedly B. Sinus node function and conduction system before and after surgery for secundum atrial septal defect: an electrophysiologic study. Am J Cardiol 1984; 53: 1415-1450.
16. Karpawich PP, Antillon JR, Cappola PR, Agarwal KC. Pre- and postoperative electrophysiologic assessment of children with secundum atrial septal defect. Am J Cardiol 1985; 55: 519-521.
17. Berdahl LD, Wenstrom KD, Hanson JW. Web neck anomaly and its association with congenital heart disease. Am J Med Genet 1995; 56: 304-307.