—Report on Experiments and Clinical Cases—

A Case Report of Spondyloepiphyseal Dysplasia Congenita

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Abstract

Spondyloepiphyseal dysplasia congenita (SED) is a rare form of skeletal systemic disease, characterized by congenital dwarfism with a short trunk and epiphysial dysplasia in the long bones and vertebral bodies. Patients also frequently saffer from atlanto-axial instability due to os odontoideum. Compression of the spinal cord caused by atlanto-axial instability is a common, serious complication in SED patients, and causes severe spinal cord symptoms or occasionally sudden death. We present an SED patient who underwent a posterior fusion of the occiput to the cervical spine for severe spinal cord symptoms due to atlanto-axial instability. (J Nippon Med Sch 2001; 68: 186—189)

Key words: dwarfism with a short trunk, paresis of the lower extremities, atlanto-axial instability, fusion operation

Introduction

Spondyloepiphyseal dysplasia congenita (SED) is a rare form of skeletal systemic disease[1], characterized by congenital dwarfism with a short trunk and epiphysial dysplasia in the long bones and vertebral bodies. In earlier studies, this disease was confused with Morquio's disease. In 1970, Spranger and Wiedemann[2-3] established the disease concept with their report on the clinical and radiographic features of 29 cases.

Since SED is frequently accompanied by atlanto-axial instability due to os odontoideum[3], which causes severe spinal cord symptoms or occasionally sudden death, its prognosis and neurological recovery depend on early diagnosis and the timing of fusion and decompression procedures.

We present an SED patient who underwent a posterior fusion of the occiput to the cervical spine for severe spinal cord symptoms due to atlanto-axial instability.

Case Presentation

A 56-year man felt numbness in both upper and lower extremities and disturbance of precise finger exercise 5 to 6 years before presentation. He was very small in stature from birth. These symptoms became gradually aggravated with the years, and on December 29, 1999, he had muscular weakness in lower extremities after falling down at home. Subsequently, he had a paresis of lower extremities and visited our clinic.

He was 96 cm tall and weighed 25 kg, indicating dwarfism with a short trunk. Neurological examination revealed hypesthesia below C 6, hyperactive deep reflex in both limbs, positive Babinski reflex, muscle weakness below the biceps brachii and disturbance of precise finger exercise. No mental retarda-

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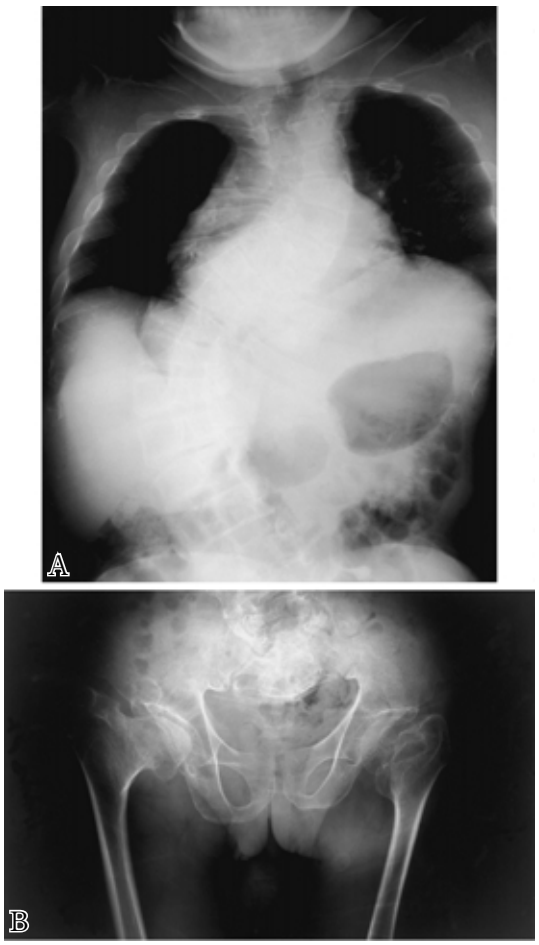

Fig. 1 Radiographic findings of the spine and the hip joint. A: flat vertebral bodies with severe scoliosis, B: proximal epiphysis dysplasia of the femur and the coxa varus.

tion was observed. Radiographic examination displayed proximal epiphysial dysplasia of the femur, the coxa varus and flat vertebral bodies with severe scoliosis (**Fig. 1**). Tomography demonstrated atlantoaxial instability due to os odontoideum associated with a narrow spinal canal at C 1 level (11 mm in sagittal diameter) (**Fig. 2**). Magnetic resonance imaging revealed a severe degree of spinal cord compression at C 1–2 level and high signal legion in the cord in a T 2-weighted image (**Fig. 3**). Percent ventilation capacity was 39.4%, indicating a high degree of expiration disturbance, possibly due to a prominent deformity of the thorax. The measurement of glycosaminoglycans in the urine was performed by the dimethylmethylene blue method for the differential diagnosis from Morquio's disease as mucopolysaccharidosis, which showed a markedly high level of haparan sulfate. The value of haparan sulfate was 2.3 mg/mmol creatinine,

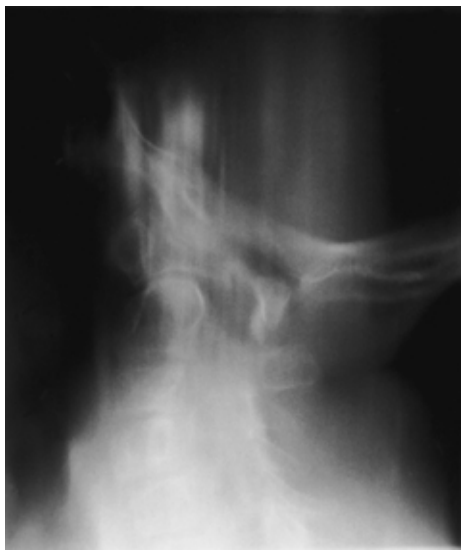

Fig. 2 Tomography of the cervical spine. Atlantoaxial instability due to os odontoideum associated with a narrow spinal canal at C 1 level (11 mm in sagtital diameter) was shown.

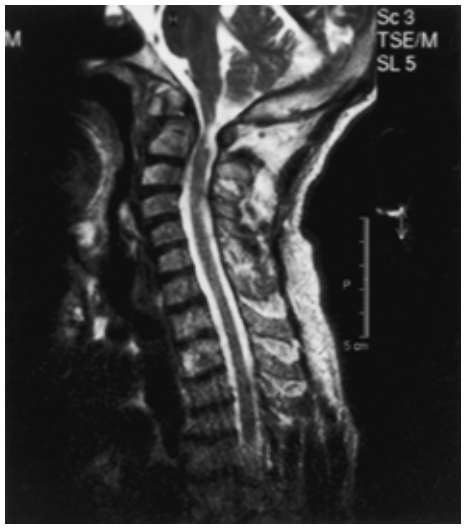

Fig. 3 Magnetic resonance imaging. Magnetic resonance imaging revealed severe degree of spinal cord compression at C 1–2 level and high signal legion in the cord in T 2-weighted image.

which is within the normal range. The diagnosis of SED was made based on dwarfism with a short trunk, characteristic radiographic features and normal urinal level of glycosaminoglycans.

On February 8, 2000, laminectomy of C 1 and posterior fusion of the occiput to the cervical spine using an SS-rod (Ransford model, child type) and iliac bone graft were done. Postoperatively, neural impairments

gradually improved with solid bone fusion (**Fig. 4**). At follow-up 8 months later the patient could walk with a walker, but still had some neurological impairment in the limbs (**Fig. 5**).

Discussion

Well over a hundred different kinds of skeletal dysplasia have been described. SED is a skeletal systemic disease accompanied by dwarfism with a short trunk, hypoplasia of the femoral head, coxa vara, flat vertebral bodies and scoliosis. As candidates for differential diagnosis, Morqio's disease, Dyggyve-Melchior-Clausen dysplasia and Kniest-Stickler dysplasia are important. The characteristics of clinical and radiographic findings in these diseases are shown in **Table 1**.

In previous reports concerning SED, os odontoideum was observed in almost all patients, and atlanto-axial instability was found in with 35—60% of patients. The onset of the occurrence of spinal cord symptom varied from childhood to over 40 years of age among those investigated. This variation has been thought to relate to the extents of atlanto-axial instability and spinal canal stenosis. In our review of recent reports of 11 SED patients treated by surgical interventions[5-12], 3 child patients had no past history of injuries. In contrast, in 5 of 8 adult patients (62.5%), spi-

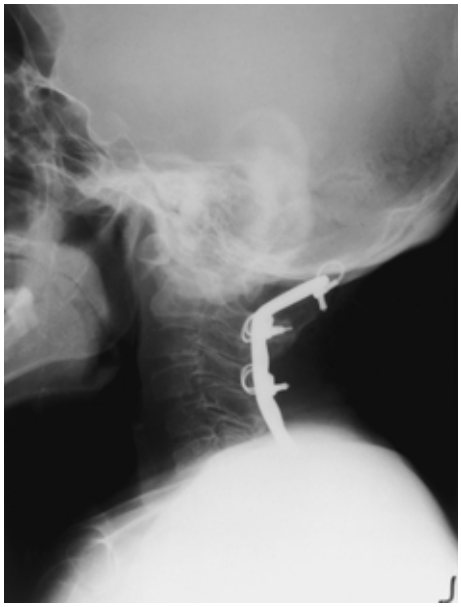

Fig. 4 Postoperative radiography of the cervical spine

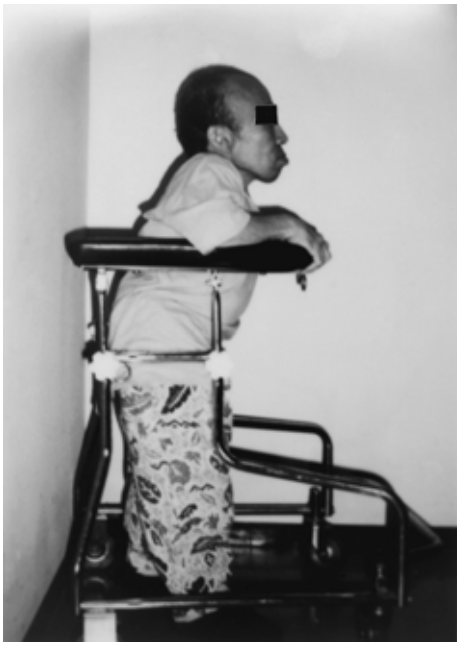

Fig. 5 A picture of the patient at follow-up 8 months later. He could walk with a walker, but still had some neurological impairment in the limbs.

Table 1 Differential diagnosis

	SED	Morquio	DMCD	KSD
Clincal findings				
Mental retardation	−	+	+	−
Cloudiness of the cornea	−	+	−	+
Joint contracture	−	−	+	+
Radiographic findings				
Vertebral body	flat body	tongue protrusion	endoplate notch	flat body
Pelvis	iliac flaring	severe iliac flaring	lace-like border	trefoil pelvis
Hip joint	coxa vara	coxa valgus	coxa vara	coxa vara
Finger	—	BLD	BLD	BLD

SED: spondyloepiphyseal dysplasia congenita, DMCD: Dyggyve-Melchior-Clausen dysplasia, KSD: Kniest-Stickler dysplasia, BLD: bullet-like deformity.

Table 2 Previous reports concerning SED patients with surgical treatment

Author	year	age	sex	causative injury
Shiino	1991	11	F	−
Saga	1997	5	M	−
Takeda	1997	11	F	−
Takada	1986	32	F	+
		24	F	+
Nii	1993	37	F	+
Uehara	1995	34	M	−
		28	M	+
Yoshino	1995	38	M	+
Yamazaki	1999	49	F	−
		32	M	−

nal cord symptoms became manifest following injuries, and patients with a history of injuries showed poor neurological recovery, compared with those without injuries (**Table 2**). Thus, patients, especially adult patients with atlanto-axial instability, may require some preventive surgical management before the occurrence of traumatic spinal damage.

Nakamura et al[13] reported that all SED patients with spinal canal diameter of 10 mm or less in the sagittal plane developed spinal cord symptoms. Abe et al[14] proposed the instability index as a risk factor against the occurrence of spinal cord symptoms: instability index = (maximal diameter of the spinal canal − minimal diameter of the spinal canal)/maximal diameter of the spinal canal × 100. They noted that surgical treatment is indicated, regardless of spinal cord symptoms, if the instability index exceeds 20%, or if the maximal diameter of the spinal canal is 15 mm or less. Compression of the spinal cord caused by atlanto-axial instability is a common, serious complication in SED patients. Since the benefit of surgery is largely prophylactic in our opinion, the absence of spinal cord symptoms does not weaken indications for the need of fusion and decompression operations.

References

1. Spranger JW, Wiedemann HR: Dysplasia Spondyloepiphysaria congenita. Helvet paediat acta 1966; 21: 598–611 (in German).
2. Spranger JW, Langer LO: Spondyloepiphyseal dysplasia congenita. Radiology 1970; 94: 313–322.
3. Ikegawa S: Spondyloepiphyseal dysplasia congenital. Shouninaika 1998; 30: 294–297 (in Japanese).
4. Spranger JW, Wiedemann HR: Dysplasia spondyloepiphysaria congenita. Helv. Joung JGN, Wevers RA, ambeek RL: Measuring urinary glycosaminoglycans in the presence of protein: An improved screening procedure for mucopolysaccharidoses based on dimethylmethylene-blue. Clin Chem 1992; 38: 803–807.
5. Svensson O, Aaro S: Cervical instability in skeletal dysplasia. Acta Orthop Scand 1988; 59: 66–70.
6. Saga I, Yoshida H, Kurihashi A, Saotome K: Infantile spondyloepiphyseal dysplasia complicating atlantoaxial subluxation. Seikeisaigaigeka 1997; 40: 403–408 (in Japanese).
7. Takeda E, Hashimoto T, Tayama M, Miyazaki M, Shirakawa E, Shiino Y, Saijo T, Ito M, Naito E, Mahbubul AHM, Kuroda Y: Diagnosis of atlantoaxial subluxation in Morquio's syndrome and spondyloepiphyseal dysplasia congenita. Acta Paediatr Jpn 1991; 33: 633–638.
8. Takada K, Igata T, Kato N, Okada Y, Mori S, Kato S, Abe H: Mother and her two children of spondyloepiphyseal dysplasia congenita with os odontoideum. Seikeigeka 1986; 37: 695–706 (in Japanese).
9. Uehara K, Sato S, Kinjo Y, Yara T, Isa S, Tanahara Y, Ibaraki K, Takara H: Two surgical cases of os odontoideum in spondyloepiphyseal dysplasia tarda. Seikeigekatosaigaigeka 1995; 44: 535–540 (in Japanese).
10. Nii E, Obokata K, Hara C, Shiokawa Y, Sudo A, Ogihara Y, Yamazaki M: Three cases of spondyloepiphyseal dysplasia congenita. Seikeigwka 1993; 44: 949–955 (in Japanese).
11. Yoshino H, Nakayama Y, Shimizu K, Yamamuro T, Yamanaka C: A case of spondyloepiphyseal dysplasia congenita with basilar impression. Rinshouseikeigeka 1995; 30: 79–81 (in Japanese).
12. Yamazaki D, Morio Y, Nagashima H, Yamamoto K: Two cases of spondyloepiphyseal dysplasia congenita accompanied by hypoplasia of odontoid process. Seikeigekatosaigaigeka 1999; 48: 61–66 (in Japanese).
13. Nakamura K, Miyoshi K, Haga N, Kurokawa T: Risk factors of myelopathy at the atlantoaxial level in spondyloepiphyseal dysplasia congenita. Arch Orthop Trauma Surg 1998; 117: 468–470.
14. Abe H, Tsuru M, Mitsumori K, Tsunoda M, Takagi H: Atlanto-axial dislocation-instability index and indications for surgery. Noushinkeigeka 1976; 4 : 57–72 (in Japanese).

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