Spinal Dysraphism: A Cross-sectional and Retrospective Multidisciplinary Clinic-based Study

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Background: Spinal dysraphism is a common birth defect that causes different kinds of secondary impairments, including joint deformities, reduced mobility, and bowel/bladder dysfunction. Due to the diversity in terminology, cultural/ethnic differences, and medical policies, prior study results cannot be generalized to all populations. Therefore, we performed this study to define the characteristics of patients in Taiwan with spinal dysraphism.

Methods: Patients diagnosed with a myelomeningocele or lipomyelomeningocele were identified from the database of our spinal dysraphism multidisciplinary clinic. A cross-sectional study was conducted by telephone interview and retrospective chart review. Clinical characteristics, such as neurologic level, orthopedic deformities, assistive device use, and level of ambulation, were collected. Spearman's correlation (*r*) tests were performed between ambulation or neurologic level and other variables.

Results: Seventy-eight subjects were included in the current study. Subjects with myelomeningoceles had more severe neurologic involvement, poorer ambulation outcome, and higher rates of orthopedic deformities, assistive device use, lower hand function, and bowel/bladder dysfunction. The correlation test revealed that the level of ambulation was negatively influenced by a higher neurologic level, a history of shunt placement, and various orthopedic deformities. Neurologic level also had widespread influence on history of shunt placement, orthopedic deformities, assistive device use, the need for additional assistive devices, aggressiveness of assistive devices, and bowel/bladder dysfunction.

Conclusion: For patients with spinal dysraphism, the neurologic level is the most important prognostic factor for many other clinical characteristics, including ambulation status. [*J Chin Med Assoc* 2008;71(10):502–508]

Key Words: ambulation, assistive devices, neurologic deficits, spinal dysraphism

Introduction

Spinal dysraphism (SD) includes the overall group of defects derived from the maldevelopment of the ectodermal, mesodermal, and neuroectodermal tissues. SD is categorized into open spinal dysraphism (OSD) and closed spinal dysraphism (CSD).[1] In OSD, the nervous tissue is exposed to the environment, whereas in CSD, the nervous tissue is covered by skin. The term *spina bifida* is still commonly used as a synonym for SD, although it properly refers to defective fusion of the posterior spinal bony elements.[2] The confusing terms *spina bifida aperta* or *cystica* and *spina bifida occulta* were once used to refer to OSD and CSD, respectively,[3] but have been progressively eliminated.

With advances in surgical and medical treatment, long-term care for patients with SD has become an important issue. The primary lesion of SD is in the spinal portion of the central nervous system, but secondary impairments may include joint deformities, reduced mobility, fecal or urinary incontinence, hydrocephalus, and cognitive dysfunction. Therefore, multidisciplinary clinical follow-up involving professionals, such as pediatric neurosurgeons, rehabilitation physicians, nephrologists,

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urologists, orthopedists, nurses, physical therapists, and orthotists, is highly advocated.

Most studies have focused on patients with myelomeningocele (MMC), which comprises the overwhelming number of patients with OSD. Few reports have been published on patients with CSD,[4–7] in which lipomyelomeningocele (LMMC) is the most common subtype.[7,8] MMC and LMMC have similar clinical presentations, except some kinds of central nervous system involvement are rare among patients with LMMC.[7,8]

Due to the diversity in terminology, cultural/ethnic differences, and medical policies, prior study results cannot be generalized to all populations. The aim of this study was to describe the clinical features and their correlation within patients in Taiwan with SD as a clinical guide for future follow-up and management.

Methods

A cross-sectional study was conducted by telephone interview among patients diagnosed with MMC or LMMC since 1981 in our single center-based SD multidisciplinary clinic database. The semi-structured telephone interview was performed by one of the authors and data on symptomatic Arnold Chiari type 2 malformations (ACM2), a history of ventriculoperitoneal shunting, scoliosis, foot deformities, hip problems (including hip subluxation, dislocation, and surgery), use of assistive devices, wheelchair use, use of lower extremity orthotics or walking aids, the need for additional assistive devices, complications from assistive device use, hand function, bowel function, bladder function, and ambulation status were collected. In addition, retrospective chart reviews were conducted and the following data was collected: age, gender, family history, pregnancy history, prenatal or postnatal diagnosis, mode of delivery, and neurologic level (S2 below, sacral; L5/S1, lumbosacral; L3/L4, mid-lumbar; L1/L2, high lumbar; thoracic). We also ranked assistive device use (from the least to the most aggressive) for wheelchairs (no wheelchair, manual wheelchair, powered wheelchair), lower-extremity orthotics (none, insole or soft ankle brace, foot orthosis, ankle-foot orthosis, knee-ankle-foot orthosis, hip-knee-ankle-foot orthosis), and walking aids (none, cane, walker used mostly indoors, forearm crutch, axillary crutch).

We performed both descriptive and statistical analyses. All statistical analyses were conducted with SPSS version 15.0 (SPSS Inc., Chicago, IL, USA). A p value of 0.05 was set as indicative of statistical significance.

Results

There were 91 and 99 patients diagnosed with MMC and LMMC in the database, respectively. We completed telephone interviews and chart reviews for 39 patients in each group; thus, 78 patients were included in this study. The reasons some patients were not included were incorrect telephone numbers (102 patients), loss of chart records (9 patients), and death (1 patient).

General data and the clinical characteristics of our subjects are listed in Table 1. Pregnancy and family histories were without clinical significance and so are not listed. The median age of the patients was 15.24 ± 6.74 years, and the percentage of male subjects was 57.7%. The overall cesarean section rate was 42.1%.

Only subjects with MMC had symptomatic ACM2 or a history of shunt placement and had been reported to have lower hand function (significantly lower dexterity or muscle power compared with peers) or were diagnosed prenatally. Subjects with MMC had higher rates of scoliosis, foot deformities, hip problems, use of assistive devices, wheelchairs, lower-extremity orthotics, or walking aids, the need for additional assistive devices, complications from assistive device use, and bowel/bladder dysfunction than subjects with LMMC.

The distribution of neurologic and ambulation levels is listed in Table 2. Five (12.8%) subjects with MMC and 18 (46.2%) with LMMC had no neurologic deficits. No subjects with LMMC had a high neurologic level (high lumbar plus thoracic). Neurologic involvement was less severe in subjects with LMMC than in subjects with MMC.

Most subjects (87.2%) with LMMC were community ambulators, and approximately half of the subjects (56.4%) with MMC were within this ambulatory status (Table 2). Eleven subjects (28.2%) with MMC and 3 subjects (7.7%) with LMMC were non-ambulators. Ambulation prognosis was better in subjects with LMMC.

The neurologic levels and corresponding ambulation levels and assistive device use are listed in Table 3. Most subjects (73.8%) with a low neurologic level (sacral plus lumbosacral) were community ambulators, unlike those with a high neurologic level. Ambulation status was more variable in subjects with a mid-lumbar neurologic level. Manual wheelchairs, ankle-foot orthoses and walkers were the most commonly used assistive devices.

Spearman's rank correlation of ambulation or neurologic levels and other variables is listed in Table 4. Ambulation level was significantly correlated with neurologic level, history of shunt placement, and various

Table 1. General data and clinical characteristics*

	MMC	LMMC	Total
Cases	39	39	78
Age, yr	12.85±4.65	17.64±7.65	15.24±6.74
Male:female	22:17	23:16	45:33
Pre:Post	5:31	0:36	5:67
VD:CS	19:19	25:13	44:32
Symptomatic ACM2	1 (2.6)	0 (0)	1 (1.3)
VP shunting history	24 (61.5)	0 (0)	24 (30.8)
Scoliosis	13 (33.3)	9 (23.1)	22 (28.2)
Foot deformities	27 (71.1)	22 (56.4)	49 (63.3)
Hip problems	6 (15.4)	3 (7.7)	9 (11.5)
Assistive device user	27 (69.2)	14 (35.9)	41 (52.6)
Wheelchair user	12 (30.8)	7 (17.9)	19 (24.4)
Manual wheelchair	11 (28.2)	7 (17.9)	18 (23.1)
Powered wheelchair	2 (5.1)	1 (2.6)	3 (3.8)
Lower-extremity orthosis user	22 (56.4)	11 (28.2)	33 (42.3)
Walking aid user	9 (23.1)	3 (7.7)	12 (15.4)
Need for additional device	14 (35.9)	9 (23.1)	23 (29.5)
Complication from device use	14 (35.9)	6 (15.4)	20 (25.6)
Lower hand function	7 (17.9)	0 (0)	7 (9.0)
Bowel dysfunction	37 (94.9)	18 (46.2)	55 (70.5)
Bladder dysfunction	37 (94.9)	20 (51.3)	57 (73.1)

**Data presented as n or mean±standard deviation or n (%). MMC = myelomeningocele; LMMC = lipomyelomeningocele; Pre = prenatal diagnosis; Post = postnatal diagnosis; VD = vaginal delivery; CS = cesarean section; ACM2 = Arnold Chiari type 2 malformation; VP = ventriculoperitoneal.*

Table 2. Distribution of neurologic level and ambulation level*

	LMMC	MMC	Total
Neurologic level			
No	18 (46.2)	5 (12.8)	23 (29.5)
S	7 (17.9)	7 (17.9)	14 (17.9)
LS	8 (20.5)	20 (51.3)	28 (35.9)
ML	6 (15.4)	3 (7.7)	9 (11.5)
HL	0 (0)	3 (7.7)	3 (3.8)
T	0 (0)	1 (2.6)	1 (1.3)
Ambulation level			
Community	34 (87.2)	22 (56.4)	56 (71.8)
Household	2 (5.1)	6 (15.4)	8 (10.3)
Non-ambulator	3 (7.7)	11 (28.2)	14 (17.9)

**Data presented as n (%). LMMC = lipomyelomeningocele; MMC = myelomeningocele; No = no neurologic deficit; S = sacral; LS = lumbosacral; ML = midlumbar; HL = high lumbar; T = thoracic.*

orthopedic deformities. Neurologic level correlated with history of shunt placement, various orthopedic deformities, various assistive device use, need for additional assistive devices, aggressiveness of assistive devices, and bowel/bladder dysfunction.

Discussion

The prevalence of OSD has been decreasing worldwide over the past 3 decades.[9] This decline has been attributed to improved maternal nutrition, periconceptional folic

Table 3. Neurologic level and corresponding ambulation level and assistive device use

	Neurologic level (no. of subjects)					
	No	S	LS	ML	HL	T
Ambulation level						
Community	22	12	19	3	0	0
Household	0	0	5	2	0	1
Non-ambulator	1	2	4	4	3	0
Assistive device use						
Not using wheelchair	23	12	21	2	0	1
Manual wheelchair	0	2	5	7	2	0
Electric wheelchair	0	0	2	0	1	0
Total wheelchair use	0	2	7	7	3	0
Lower extremity orthosis use						
None	19	10	13	2	1	0
Insole or ankle brace	0	1	1	0	0	0
Foot orthosis	2	1	0	0	1	0
Ankle-foot orthosis	2	1	12	6	1	0
Knee-ankle-foot orthosis	0	0	1	1	0	0
Hip-knee-ankle-foot orthosis	0	1	1	0	0	1
Total lower extremity orthosis use	4	4	15	7	2	1
Walking aid use						
None	23	13	20	6	3	1
Cane	0	0	2	0	0	0
Walker	0	1	4	1	0	0
Forearm crutch	0	0	1	1	0	0
Axillary crutch	0	0	1	1	0	0
Total walking aid use	0	1	8	3	0	0

No = no neurologic deficit; S = sacral; LS = lumbosacral; ML = midlumbar; HL = high lumbar; T = thoracic.

Table 4. Spearman's rank correlation between neurologic level or ambulation level and other variables

Neurologic level		Ambulation level	
Shunting history	0.370*	Shunting history	0.500*
Scoliosis	0.344*	Scoliosis	0.266†
Foot deformities	0.588*	Foot deformities	0.325*
Hip problems	0.310*	Hip problems	0.334*
Assistive device use	0.614*	Neurological level	0.506*
Wheelchair use	0.538*	Shunting history‡	0.505*
Lower extremity orthosis use	0.431*		
Walking aid use	0.294*		
Need for additional device	0.457*		
Bowel dysfunction	0.597*		
Bladder dysfunction	0.589*		
Wheelchair aggressiveness	0.535*		
Lower extremity orthosis aggressiveness	0.464*		
Walking aid aggressiveness	0.299*		
Shunting history‡	0.348†		

**$p < 0.01$; †$p < 0.05$; ‡correlation was performed within the myelomeningocele group.*

acid supplementation, wider availability of prenatal diagnostic tests with an increased incidence of elective terminations, and other unknown reasons.[10] However, SD is still the second most common congenital birth defect worldwide.[11] MMC remains the most common congenital neurological malformation compatible with life.[10]

MMC and LMMC have the same clinical presentation in many aspects, such as lower extremity paralysis,

sensory loss, musculoskeletal deformity, and neurogenic bladder. Nevertheless, hydrocephalus and ACM2 are generally not associated with LMMC.[7,8] These observations concur with our finding that no subjects with LMMC had symptomatic ACM2, shunts placed to resolve hydrocephalus, or lower hand function which might have resulted from hydrocephalus or ACM2.

Only 1 child (2.6%) with MMC had history of symptomatic ACM2. The reason for this low prevalence compared to that in another study[12] with a prevalence of up to 20% might be that not all of the subjects had been followed-up since birth in our clinic. Symptomatic ACM2 often occurs in the first few years of life and has a relatively high mortality. Furthermore, the study group comprised only 41.1% (78/190) of the original sample. Lastly, there was poor comprehension of ACM2 by the parents.

No subjects with LMMC were diagnosed prenatally, compared to 5 (13.9%) subjects with MMC. Prenatal diagnosis of MMC is now routinely performed by obtaining maternal serum α-fetoprotein and ultrasound followed by amniocentesis or detailed sonography, if indicated. In contrast, no reliable prenatal diagnostic methods for LMMC exist. Only a few case reports[13–15] have described the prenatal sonographic detection of LMMC.

Patients and their families are often most concerned at the time of diagnosis about the prognosis for ambulation. The ambulation level was determined according to Hoffer et al's classification[16] in our study, but therapeutic (nonfunctional) ambulators and non-ambulators were combined to the same level, since nearly all therapeutic ambulators will become non-ambulators over time.[17,18] The Sharrard classification[19] was used for the neurologic level. In patients with MMC, neurologic level was considered to be the determining factor influencing ambulation.[17,20–22] This general agreement also applies to patients with other kinds of SD, including LMMC,[5] and was found in our study as well, with a high Spearman's rank correlation between neurologic and ambulation levels. This also explains that subjects with MMC had more severe neurologic involvement as well as poorer ambulation outcome compared to subjects with LMMC.

In addition to the neurologic level, there are additional prognostic factors, including upper motor neuron signs such as spasticity, tethered cord, syringohydromyelia, ACM2,[18,23,24] type of bracing,[25,26] musculoskeletal deformities,[16,17] changing social goals, larger body mass due to maturation,[27] a period of immobilization,[28] and even poor motivation.[29] Such additional prognostic factors could explain why some subjects with a low neurologic level would have an unexpectedly worse ambulation outcome. One extreme example was a patient in our study without neurologic deficit, defined by muscle power of the lower extremities, who was a non-ambulator (Table 3). He had a symptomatic ACM2 and had received surgery. The sequelae, including spasticity, cognitive dysfunction, possibly poor balance, and poor muscle control, made him a non-ambulator. This reveals the drawback of using neurologic level as a single prognostic factor for ambulation.

In our study, scoliosis, foot deformities, and hip problems had a negative impact on the subject's ambulation status. This was in agreement with previous studies.[16,17] Shunting history also exacerbated subjects' ambulation status in our study. Since only subjects with MMC would have a history of shunt placement, we analyzed the correlation between ambulation and shunting history within the MMC group. The result also revealed the same negative impact. Some studies[29,30] have suggested that multiple shunt revisions is a risk factor for a poorer ambulation level. We did not collect data regarding shunt revision, only shunting history. Therefore, further studies are needed to clarify the influence of shunt revision on the ambulation status of our patients.

In addition to ambulation, neurologic level also had a significant influence on many other variables in our study, as we had anticipated. Specifically, the higher/worse the neurologic level, the higher the prevalence of shunting history, scoliosis, foot deformities, hip problems, use of assistive device, use of wheelchair, use of lower extremity orthotic, use of walking aid, need for additional devices, and bowel/bladder dysfunction. A history of shunt placement was not associated with LMMC. Therefore, we performed a correlation test between neurologic level and history of shunt placement within the MMC group. The result was also significant, and this concurred with the finding of a study[4] that had involved 142 subjects with OSD. The same study,[4] with a total of 179 subjects with OSD or CSD, also suggested that the higher the neurologic level, the higher the prevalence of scoliosis and foot deformities. Our findings, again, concurred with that. The neurologic level was also associated with the type of assistive device use, as we had anticipated. After we ranked assistive device use from the least to the most aggressive, we showed that subjects with a higher/worse neurologic level used the more aggressive wheelchairs, lower-extremity orthotics, and walking aids.

In a cross-sectional study[31] with a sample size of 348 subjects (mean age, 18 years) diagnosed with MMC, 52%, 5%, 35%, and 23% of the subjects used a manual wheelchair, powered wheelchair, lower-extremity

orthotic, and walking aid, respectively, compared to 28.2%, 5.1%, 56.4%, and 23.1% of the subjects in our MMC group, respectively (Table 1). The percentages of powered wheelchair users and walking aid users were nearly the same between the 2 studies; however, our subjects with MMC used the manual wheelchair less frequently, but used lower-extremity orthotics more often. The less frequent use of manual wheelchairs might be due to the idea of wheelchair use representing a handicap, of insufficient social welfare support for wheelchair use in our nation, poor family economic status, unrealistic ambulation goals, or other unidentified factors. More of our subjects used lower extremity orthotics than in a previous study.[31] This discrepancy is hard to explain; physician's preference, local medical policy, and parents'/patients' attitudes might be possible reasons, but these are just presumptions. The true causes and all our presumptions require clarification through further study.

Among the 41 subjects using assistive devices, 20 (48.8%) had experienced complications from device use (Table 1). Skin breakdown, pain, and falls comprised most of the complications, and all of the complications would usually cause a period of abstinence from assistive device use, immobilization, or a deterioration in ambulation status. This is a problem requiring serious treatment, by way of correct device prescription, sufficient instructions about device use, and periodic follow-up.

There were several limitations to our study. The first was inherent in the cross-sectional nature of the study by telephone interview. A variety of information could not be convincingly collected by telephone interview. We did not examine the patients directly, and the current status of spasticity, contractures, scoliosis, foot deformities, hip dislocations, other orthopedic deformities, and the assistive device design were unclear. The prevalence might have been under-reported or even over-reported for various reasons. The second limitation was that many subjects in the original sample were not included in the study sample. This might have had a significant influence, although we assumed that the incidence of having an incorrect telephone number, the main reason for the subjects' exclusion, should have been distributed randomly throughout the original sample. However, this was just our presumption. This limitation also resulted in a relatively small study sample size (78 subjects).

Even though there were limitations and presumptions within our study, we believe that this study can serve as a background for further study designs, especially prospective studies with or without focusing on a specific issue.

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