

## MALFORMATIONS OF THE INTERNAL ORGANS AND SYSTEMS IN CHILDREN WITH ASYMPTOMATIC SPINAL DYSRAPHISM

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**Purpose.** We determined the incidence of associated malformations in children with latent forms of spinal dysraphism.

**Materials and methods.** The study included 64 children aged 9 months to 17 years with latent forms of spinal dysraphism. We evaluated the spine and spinal canal states and orthopedic and neurological statuses on the basis of clinical, radiological, and magnetic resonance imaging findings.

**Results.** Malformations of the spine were observed in 100% of the patients, and associated malformations of the organs and systems were found in 33 (52%) patients. Malformations of the genitourinary, musculoskeletal, cardiovascular, digestive, otolaryngological, and bronchopulmonary systems were observed in 52%, 45%, 39%, 12%, 9%, and 3% of the patients, respectively.

**Conclusion.** Pediatric patients with latent forms of spinal dysraphism require detailed examinations of the spine, spinal canal, and internal organs and systems. The most frequently occurring malformations were in the genitourinary, musculoskeletal (appendicular skeleton), and cardiovascular systems.

**Keywords:** children, latent spinal dysraphism, congenital malformations of the spine, abnormal internal organs.

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### Introduction

Abnormalities of the spinal cord and spinal canal, known as spinal dysraphisms, include a group of congenital conditions, different in form but united by a common mechanism of development; specifically, incomplete fusion of the midline embryonic mesenchymal bone and nerve structures and accompanied by obligatory involvement of the spinal cord in the pathological process [1]. According to the literature, the incidence of spinal dysraphism is 0.5–2.5 per 1,000 live births, depending on race, ethnicity, and region of residence. Defects form during different stages of embryogenesis (gastrulation, primary, and secondary neurulation) [2]. Spinal dysraphisms are conventionally divided into open and occult forms. By definition, occult forms of spinal dysraphism are characterized by maintaining the integrity of the overlying skin and may be combined with cutaneous stigmata located directly in the area of dysraphia or at some distance from it [3]. The course and prognosis of occult forms of spinal dysraphism

depend on the severity of clinical symptoms and the presence of comorbidity with other organs and systems. The clinical manifestations of occult forms of spinal dysraphism include cutaneous stigmata, bone and joint deformities, defects of musculoskeletal system functions, neurological disorders of varying severity, and disorders of the pelvic organs. In the literature, there are a few reported studies on the co-incidence of spinal canal pathologies and developmental abnormalities of the internal organs in children with congenital deformities of the spine [4–12]. However, we have not seen reports specifically devoted to the co-incidence of birth defects of the spine and malformations of other organs and systems of children with occult spinal dysraphism.

The purpose of this study was to determine the types and frequencies of occurrence of malformations of the spine and of the internal organs and systems in children with occult spinal dysraphism.

## Material and Methods

From 2007 to 2014, in the clinic of Spine and Neurosurgery of the Turner Scientific and Research Institute for Children's Orthopedics, Russian Ministry of Health, 64 children (15 boys and 49 girls) between the ages of 9 months and 17 years were diagnosed with occult spinal dysraphism. All children were examined clinically and using diagnostic instruments. During the clinical examinations, we evaluated the orthopedic and neurological statuses of the patients. X-ray examination, including spondylography of the frontal and lateral projections, multislice computed tomography (CT) of the spine, and magnetic resonance imaging (MRI) of the thoracic and lumbosacral segments of the vertebral column, were performed. In addition, the physical examination of the patients in the hospital included ultrasound of the abdominal area, kidneys, and heart as well as examinations by pediatricians, neurologists, cardiologists, and other specialists.

## Results

In the course of physical examination, spinal deformities were observed in 52 (81%) children. S-shaped scoliosis was observed in 13 patients in the thoracic segment, in 8 children in the thoracolumbar junction, and in 10 patients in the lumbar segment. Scoliotic arches were observed in the thoracic and lumbar spine in four patients. Kyphotic deformities were observed in seven patients in the thoracic segment, in seven patients in the thoracolumbar segment, and in two patients in the lumbar segment. One patient only had kyphosis of the thoracic segment. The remaining 12 (19%) patients did not have spinal axis deviations in the frontal and sagittal plane.

Skin stigmata were observed in 23 (36%) patients. Local hypertrichosis in the area of dysraphism was noted in 12, dorsal dermal sinus was observed in 1, subcutaneous lipoma was detected in 5, and local retraction of the skin in the lumbosacral region was observed in 2 patients. Multiple cutaneous stigmata caused by defective embryogenesis were found in three patients: local and nevus hypertrichosis in one patient, dorsal dermal sinus and local hypertrichosis in one, and local skin retraction and hemangioma in one patient. No cutaneous stigmata were found in 41 (64%) patients.

Neurological deficits were detected in 45 (70%) children. Lower extremity abnormalities were detected in 44 patients, of whom 28 exhibited lower paraparesis and 16 exhibited monoparesis. Out of these patients, 26 clinically showed unilateral tibial muscle wasting in combination with shortening of the limbs due to truncated tibia in 13 and in combination with foot hypoplasia in 7. Clubfoot deformities were observed in five children (subtypes: three equinovarus, one equinus, and one varus). Pelvic organs dysfunctions were observed in 10 (16%) of 64 patients, 9 of whom also had lower mono- and paraparesis. Neurological deficits were absent in 19 (30%) patients.

The distribution of forms of occult spinal dysraphism in the patients was as follows. In 49 (77%) patients, occult spinal dysraphism was present as an isolated defect: diastematomyelia type 1 in 34 patients, filum terminale lipoma in 11, intradural spinal lipoma in 2, diastematomyelia type 2 in 1, and dorsal dermal sinus in 1. In 15 (23%) patients, there was a combination of occult forms of spinal dysraphia: diastematomyelia type 1 was combined with filum terminale lipoma in eight patients and with intradural spinal lipoma in four patients, and filum terminale lipoma was combined with dorsal dermal sinus in one patient. Diastematomyelia type 2 was combined with filum terminale lipoma in two patients.

As shown in Table 1, X-ray examination showed vertebral malformation in 5 (8%), vertebral segmentation defects in 14 (22%), vertebral fusion defects in 5 (8%) patients, and combined anomalies of the vertebrae in 40 (62%) children. Localized anomalies of the vertebrae of the thoracic segment were observed in 26 patients, of the lumbar segment in 12, and of both the thoracic and lumbar segments in 26 patients. Concrescence of the ribs in conjunction with vertebral fusion defects was observed in 2 patients, with segmentation defects in 2 patients, and with combined anomalies of the spine in 12 patients.

In the course of physical examination, related anomalies of the internal organs and systems were found in 33 (52%) out of 64 children with occult spinal dysraphism. Genitourinary defects were detected in 52% of the patients, musculoskeletal in 45%, cardiovascular system defects in 39%, digestive system defects in 12%, ear/nose/throat (ENT) in 9%, and pulmonary system defects in 3% (Table 2).

Table 1

Distribution of patients by type of spinal development defect and by sex

Type of defect	Sex	
	Males	Females
<b>1. Defects of vertebral development</b>	<b>2</b>	<b>3</b>
a. Posterolateral hemivertebrae	2	3
– single	0	3
– multiple	2	0
<b>2. Vertebral segmentation defects</b>	<b>2</b>	<b>12</b>
<b>3. Vertebral fusion defects,</b>	<b>2</b>	<b>3</b>
Butterfly vertebrae		
– single	1	2
– multiple	1	1
<b>4. Combined anomalies</b>	<b>9</b>	<b>31</b>
a. Defects of development and segmentation	3	5
b. Defects of development and fusion	0	6
c. Defects of development, segmentation, and fusion	3	10
d. Defects of segmentation and fusion	3	10
<b>Total</b>	<b>15</b>	<b>49</b>

Table 2

Malformations of the organs and systems in children with congenital defects of the spine and occult spinal dysraphism

Organs and systems	Type of anomaly	Number of anomalies	Number of patients
Urogenital	Renal agenesis	4	17
	Renal hypoplasia	3	
	Duplex kidneys	11	
	Hydrocele	1	
	Meatal stenosis	1	
	Inguinal hernia	2	
	Cryptorchism	2	
	Total: 24		
Musculoskeletal	Limb anomalies	3	15
	Congenital hip dislocation	2	
	Sprengel deformity	1	
	Umbilical hernia	2	
	Foot deformities	10	
	Total: 18		
Cardiovascular	Patent foramen ovale	3	13
	Mixed aortic defect	1	
	Left ventricular false tendons	9	
	Ventricular septal defect	1	
	Pulmonary valve stenosis	1	
	Total: 15		
Digestive	Accessory liver lobe	1	4
	Aplasia of the gallbladder	1	
	Recto-urethral fistula	1	
	Anal atresia	2	
	Total: 5		
ENT (and vision Coloboma)	Auditory canal atresia	1	3
	Microtia	2	
	Macrostomia	1	
	Coloboma	1	
	Total: 5		
Respiratory/Pulmonary	Tracheobronchmalacia	1	1
	Total: 1		

In these children with occult spinal dysraphism, 73% had type 1 diastematomyelia. Congenital malformations of the vertebrae were observed in 100% of the patients. Thus, most of the patients had a combination of vertebral anomalies (40 children). Malformations of other organs and systems were observed in 51% of the patients. The most common defects were of the urogenital tract, followed by defects in the musculoskeletal and cardiovascular systems.

## Discussion

Intradural spinal pathologies and anomalies of organs and systems in children with congenital malformations of the spine occur with varying frequency in previous reports. According to McMaster, intradural myelography revealed pathologies in 46 (18%) of 251 patients with congenital scoliosis [4]. Using MRI, Bradford et al. found pathologies of the spinal canal in 38% of 42 patients with congenital spinal deformities [5], and Suh et al. found congenital malformations of the vertebrae in 31% of 41 examined patients [6]. Shen et al. performed MRI and showed intradural pathologies in 43% of 226 children with congenital spinal deformities [7]. According to Ulrich E.V., a comprehensive survey of 223 children with various vertebral defects demonstrated the presence of associated malformations in 170 (76.2%) patients [8]. Beals et al. thoroughly examined 218 patients ranging in age from 1 to 28 years with congenital scoliosis. The authors established that 133 (61%) patients had anomalies of the internal organs and systems, including the kidneys in 8% of patients and the heart in 7% [9]. Other researchers examined 195 children with congenital spinal deformities. In 89 (46%) of the patients, anomalies were present in the genitourinary system in 76% of patients, in the musculoskeletal system in 45%, in the central nervous system in 44%, in the gastrointestinal tract in 17%, in the heart in 8%, in the ENT organs in 7%, and in the pulmonary system in 6% [10]. Shen et al. examined 226 children with congenital spinal deformities and detected anomalies of the following types in 91 (40%) of the patients: heart in 18%, genitourinary system in 12%, and gastrointestinal tract in 5%. The same researchers noted that the presence of intradural pathologies in patients with congenital spinal deformities had no effect on the incidence of malformations of the

internal organs [7]. Bollini surveyed 75 children and found comorbidities of various organs and system anomalies in 34 (45%) patients. In this group, pathologies of the genitourinary system were present in 18 (24%) patients, of the heart in 6 (8%), and of the spine in 11 (15%) [12]. Thus, the findings of the above authors and our study show similar frequencies of occurrence of malformations of the internal organs and systems in children with occult spinal dysraphism combined with congenital defects of the spine. These data suggest a notably high frequency of occurrence of occult spinal dysraphia in patients with congenital malformations of the axial skeleton, a finding that requires more detailed and in-depth examinations of patients to exclude intradural spinal pathologies.

## Conclusion

Child patients with occult forms of spinal dysraphism require detailed physical examinations of the spine and spinal canal and of the internal organs and systems. In these patients, malformations of the spinal column were observed in 100% of the patients, and more than 50% of the patients had associated anomalies of the internal organs and systems. The most common malformations were of the genitourinary, musculoskeletal, and cardiovascular systems. The high incidence of abnormalities of internal organs and systems in patients with occult forms of spinal dysraphism aggravates the condition of these patients and requires treatment during preoperative preparation.

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