Myelodysplasia in children with anorectal malformations
Mielodisplasia em crianças com malformações anorretais

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ABSTRACT

Objective: To compare computed tomography and magnetic resonance imaging accuracy to diagnose myelodysplasia in patients with anorectal anomaly, to verify the prevalence of tethered spinal cord, and to investigate if there is a direct relation between the presence of myelodysplasia and type of anorectal anomaly, bone alteration, functional prognosis and associated malformations in these patients. Methods: Thirty five patients with anorectal anomaly were studied using computed tomography and magnetic resonance imaging of lumbosacral spine. Myelodysplasia was analyzed in terms of spinal computed tomography findings, type of anorectal anomaly, sacral alterations, presence of associated anomalies and fecal and urinary continence. Results: Magnetic resonance imaging showed myelodysplasia in 45% of patients, and computed tomography provided correct diagnosis of the spinal condition in only 66% of subjects. Myelodysplasia was observed even in low anorectal anomalies. No association was found between myelodysplasia in magnetic resonance imaging and presence of vertebral alterations; associated anomalies; fecal and urinary continence. Prevalence of tethered spinal cord was of 14%. Conclusions: The best exam to diagnose myelodysplasia is magnetic resonance imaging. Vertebral malformations, urinary and fecal continence and presence of associated anomalies were not good indicators of myelodysplasia, but they could indicate tethered spinal cord.

Keywords: Rectum/anomalities; Anal canal/anomalities; Myelodysplastic syndromes/radiography; Tomography, X-ray computed; Magnetic resonance imaging

INTRODUCTION

Patients with anorectal anomaly present a high incidence of associated malformations. Alterations of the spinal cord are present in 13% to 50% (1-4), and are more frequent in the lumbosacral spine. Sacral agenesis and failure of fusion of the posterior arch are the most common vertebral alterations (3,5).

Malformation of sacral vertebrae might be associated with hypoplasia of the levator ani muscle (6-7), functional deficiency of the anal sphincter (7-10) and damage of nerve roots. These alterations might impair fecal and urinary continence (8).
Nerve alterations due to malformation of the lumbosacral spine have always been regarded as static and non-progressive. However, some patients showed progressive degeneration of their neurological condition. The development computer tomography (CT) and magnetic resonance imaging (MRI) techniques enabled establishing a direct relation between bone malformations and spinal dysraphism or myelodysplasia (MD) - a syndrome resulting from the defective median fusion of bone, mesenchyma and nerve structures(11).

Myelodysplasias include syringomyelia, diastematomyelia, lipoma of the filum terminale (FT), fatty infiltration of FT, meningocele, myelomeningocele and neuroenteric cyst. All these conditions might be associated with tethered cord - when the spinal cord is still attached to the most distal portion of the spine, therefore hindering the cephalad migration during bone growth. This stretching of the final portion of the spinal cord causes vascular insufficiency, which in turn results in neurological alterations(11-22).

Because computer tomography (CT) is frequently used at our hospital, for a long time we used CT to determine the presence of MD in patients with anorectal anomalies. However, greater access to nuclear magnetic resonance (MRI) techniques revealed a clear difference in results comparing both methods MRI.

**OBJECTIVE**

The aim of this study is to compare the accuracy of CT and MRI techniques in the diagnosis of MD and to determine the prevalence of tethered cord. We also investigated a possible direct relation between MD and the type of anorectal malformation; the presence of vertebral alterations; functional prognosis of patients, and presence of other associated malformations.

**METHODS**

We conducted a retrospective study evaluating 154 patients with anorectal malformation treated at the Pediatric Surgery Service of Hospital Infantil Darcy Vargas, between 1990 and 2005.

We included in this study 35 patients (15 female and 20 male) who underwent computer tomography (CT) scans and nuclear magnetic resonance (NMR) of the lumbosacral spine.

The diagnosis of myelodysplasia (MD) by MRI was assessed in relation to:

1. computer tomography.
2. type of anorectal malformation.

We determined the incidence of MD for each type of anorectal malformation. We used the classification described by Peña(23)

3. evaluation of the spine.

We analyzed the presence of spinal bone alterations diagnosed by CT and compared the data to the presence of MD;

4. associated malformations.

We determined the presence of heart, genitourinary and lower limb malformations and correlated to the presence of MD;

5. fecal continence.

Patients were classified as to fecal incontinence as described by Martins(24):

- grade I: complete fecal continence (up to two solid evacuations a day, with no soiling);
- grade II: partial fecal continence (3-5 times a day passing of soft stool with occasional soiling) and;
- grade III: incontinence (more than 5 evacuations a day with frequent soiling);

6. urinary continence.

Patients were considered continent if they had dry periods of at least three hours;

7. prevalence of tethered cord.

**Statistical Analysis**

The variables are presented according to absolute (n) and relative (%) frequencies.

The relation between MD diagnosis by MRI and variables of interest was assessed using the chi-square test or Fisher’s exact test.

Regarding the diagnosis of MD, we used the Kappa coefficient to compare the results between MRI and CT scans.

We considered a level of significance of 0.05 (a = 5%). The p values lower than 0.05 were considered significant and were marked with an asterisk(*).

**RESULTS**

MRI determined the presence of MD in 16 (45%) of 35 patients. Lipoma of the FT was the most common alteration (10 patients).

**CT and MRI in diagnosis of MD**

CT and MRI diagnosed the absence of MD in 18 patients. The same type of MD was diagnosed on CT and MRI in five patients. MRI diagnosed myelodysplasia in eight patients with normal CT scans. In the case of four patients, although CT scans and MRI showed the presence of MD, the results of both exams were conflicting (table 1).
Myelodysplasia in children with anorectal malformations

In our study we had several different types of anorectal malformation with low level of representation. Due to the small size of the sample we could not use statistical tests to assess the association between the type of anorectal malformation and the presence of MD.

Spinal alterations and MD

Out of five patients with no tomographic evidence of spinal bone alterations, two had MD – lipoma of the FT.

Out of 30 patients with spinal bone alterations, 13 had MD (table 4).

<table>
<thead>
<tr>
<th>Bone CT</th>
<th>MD</th>
<th>N</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>Absent</td>
<td>3</td>
</tr>
<tr>
<td>Normal</td>
<td>Lipoma of FT</td>
<td>2</td>
</tr>
<tr>
<td>Failure of fusion of the posterior arch</td>
<td>Absent</td>
<td>17</td>
</tr>
<tr>
<td>Failure of fusion of the posterior arch</td>
<td>Present</td>
<td>13</td>
</tr>
</tbody>
</table>

No statistically significant association was found between the CT diagnosis of failure of fusion of the posterior arch and the MRI diagnosis of MD ($p = 1.000$) (table 5).

In the group of patients with normal CT scans of the bones, 40% had been diagnosed with MD on MRI. In the group with bone CT diagnosis of failure of fusion of the posterior arch, 43% had MRI diagnosis of MD. The difference was not statistically significant (table 5).

<table>
<thead>
<tr>
<th>MRI - MD</th>
<th>Bone CT</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>n</td>
<td>n</td>
<td>n</td>
</tr>
<tr>
<td>Normal</td>
<td>Failure of fusion of the posterior arch</td>
<td>n</td>
</tr>
<tr>
<td>Absent</td>
<td>3 60.0</td>
<td>17 56.7</td>
</tr>
<tr>
<td>Present</td>
<td>2 40.0</td>
<td>13 43.3</td>
</tr>
<tr>
<td>Total</td>
<td>5 100.0</td>
<td>30 100.0</td>
</tr>
</tbody>
</table>

Fisher’s exact test: $p = 1.000$

Myelodysplasia and associated malformations

MRI revealed no statistically significant association between the presence of MD and the presence of associated malformations. ($p = 0.600$) (table 6).

MRI revealed the presence of MD in 41.2% of patients with associated malformations and in 50.0% of patients with no associated malformation. There was no significant difference between the two groups (table 6).
Table 6. Anomalies associated with MD

<table>
<thead>
<tr>
<th>MRI - MD</th>
<th>Associated anomalies</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>With</td>
<td>Without</td>
</tr>
<tr>
<td></td>
<td>n %</td>
<td>n %</td>
</tr>
<tr>
<td>Absent</td>
<td>10 58.8</td>
<td>9 50.0</td>
</tr>
<tr>
<td>Present</td>
<td>7 41.2</td>
<td>9 50.0</td>
</tr>
<tr>
<td>Total</td>
<td>17 100.0</td>
<td>18 100.0</td>
</tr>
</tbody>
</table>

Chi-square test: $p = 0.600$

Fecal continence

There was no statistically significant association between grade of fecal continence and presence of MD on MRI ($p = 0.136$) (table 7).

RMI revealed the presence of MD in 26.7% of patients with grade I fecal continence; in 66.7% of patients with grade II fecal continence; and in 57.1% of patients with grade III fecal continence. Despite the apparent difference between these proportions, there was no statistically significant difference between them (table 7).

Table 7. Fecal continence and MD

<table>
<thead>
<tr>
<th>MRI - MD</th>
<th>Fecal continence</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Grade I</td>
<td>Grade II</td>
</tr>
<tr>
<td></td>
<td>n %</td>
<td>n %</td>
</tr>
<tr>
<td>Absent</td>
<td>11 73.3</td>
<td>2 33.3</td>
</tr>
<tr>
<td>Present</td>
<td>4 26.7</td>
<td>4 66.7</td>
</tr>
<tr>
<td>Total</td>
<td>15 100.0</td>
<td>6 100.0</td>
</tr>
</tbody>
</table>

Chi-square test: $p = 0.136$

Urinary continence

There was no statistically significant association between urinary continence and presence of MD on MRI ($p = 0.922$) (table 8).

MRI revealed the presence of MD in 45.0% of continent patients and 46.7% of incontinent patients. No statistically significant difference was found between the two groups (table 8).

Table 8. Urinary continence and MD

<table>
<thead>
<tr>
<th>MRI - MD</th>
<th>Urinary continence</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Continent</td>
<td>Incontinent</td>
</tr>
<tr>
<td></td>
<td>n %</td>
<td>n %</td>
</tr>
<tr>
<td>Absent</td>
<td>11 55.0</td>
<td>8 53.3</td>
</tr>
<tr>
<td>Present</td>
<td>9 45.0</td>
<td>7 46.7</td>
</tr>
<tr>
<td>Total</td>
<td>20 100.0</td>
<td>15 100.0</td>
</tr>
</tbody>
</table>

Chi-square test: $p = 0.922$

Presence of tethered cord

Five (14.0%) patients had MRI diagnosis of tethered cord. Regarding the type of anomaly we noticed:
- vestibular fistula - 1;
- bulbar urethral fistula - 1;
- perineal fistula - 2.

All had associated malformations of heart and/or genitourinary system, and/or lower limbs.

Two patients had urinary continence and three were incontinent. Regarding fecal continence, one was classified as grade I, 2 as grade II and 2 as grade III. (chart 1)

Table 8. Characteristics of patients with tethered cord (TC)

<table>
<thead>
<tr>
<th>Type of anorectal malformation</th>
<th>Urinary continence</th>
<th>Fecal continence</th>
<th>Associated anomalies</th>
<th>MD</th>
</tr>
</thead>
<tbody>
<tr>
<td>F Perineal</td>
<td>Yes</td>
<td>Grade II</td>
<td>Currario syndrome</td>
<td>Syringomyelia</td>
</tr>
<tr>
<td>M Bulbar</td>
<td>No</td>
<td>Grade III</td>
<td>Hypospadia + bifid scrotum</td>
<td>Lipoma of FT</td>
</tr>
<tr>
<td>M Not defined</td>
<td>Yes</td>
<td>Grade I</td>
<td>Hypospadia</td>
<td>Lipoma of FT</td>
</tr>
<tr>
<td>M Perineal</td>
<td>No</td>
<td>Grade II</td>
<td>Vicious rotation of the left kidney</td>
<td>Lipoma of FT</td>
</tr>
<tr>
<td>F Vestibular</td>
<td>No</td>
<td>Grade III</td>
<td>Double urethra, vagina, bladder and uterus</td>
<td>Diastematomelia, syringomyelia, myelomeningocele</td>
</tr>
</tbody>
</table>

DISCUSSION

In theory, malformations of the lumbosacral spine should only have an impact on the sacral nerve roots and on development of the levator ani muscle\(^{(1)}\). Thus, alterations due to these lesions would be static and would not progress\(^{(11)}\). However, some patients presented progressive worsening of their neurological picture. This observation, together with the development of new diagnostic methods, such as CT and MRI, made it possible to determine the existence of a direct correlation between sacral malformations and MD\(^{(6)}\).

Patients with anorectal malformations present a higher incidence of spinal cord lesions, which were not suspected before. Lesions of the spinal cord in patients with anorectal malformation include tethered cord, lipoma of the FT, myelocle, myelomeningocele, diastematomyelia, syringomyelia, and a combination of these lesions\(^{(11)}\).

MD includes a number of lesions that might require surgical treatment and morbidity depends on the timing of diagnosis. MD morbidity is related to irreversible alterations of the genitourinary, gastrointestinal and skeletal muscle systems\(^{(11)}\).

In our study, 45% of patients presented MD. This result is similar to data published in previous studies where the incidence of MD was 53% and 50%\(^{(2-3)}\). Since the incidence of malformations of the lumbosacral spine
is high in patients with anorectal anomalies, and these malformations might be associated with MD, it is necessary to establish diagnostic protocols for these patients(6). An X-ray of the lumbosacral spine might reveal failure of fusion of the posterior arch, hemivertebrae, sacral agenesis, spur of diastematomyelia, and signs indicating the presence of MD. However, in pediatric patients, the radiological diagnosis (X-ray) is impaired by incomplete ossification, and by the fact that in these patients MD might be present with no evidence of bone alterations(11).

Newborns with anorectal malformation should undergo ultrasonographic screening for bone alterations and MD(25). Nevertheless, the use of ultrasound is limited to children up to six months, the maximum age for ultrasonographic diagnosis.

MRI has surpassed CT in the diagnosis of spinal cord lesions. MRI reveals anatomic and pathologic planes that are not possible with CT(12). CT is used to obtain information on muscle and bone structure, but is less specific for the diagnosis of myelodysplasias(26). In the current study, when compared to MRI, CT diagnosis of MD was correct in 23 patients. The absence of MD on CT scans did not exclude the need of MRI, since eight patients with negative MD diagnosis on CT scans presented alterations of the spinal cord on MRI.

Some authors correlated the incidence of MD with the type of anorectal malformation. The higher the incidence of MD, the higher the incidence of bone and spine alterations(10-11,20,27). In the present study, we noticed alterations in the lumbosacral spine and MD in patients with high and low anorectal anomalies. This finding underlines the need to investigate all patients regardless of the type of malformation.

Regarding bone structure evaluation with CT scans of the spine, two (40%) of five patients with no bone alterations had positive MRI diagnosis for MD. The fact that Long et al.(28) observed MD in 21% of patients with normal bone structure highlights the importance of ruling out MD even in patients with no spinal malformation.

The presence of associated cardiac, genitourinary and lower limb malformations and the presence or absence of urinary and fecal continence were not indicative of MD. Yet, associated malformations and urinary and fecal incontinence were more frequent in patients with tethered cord, which is in agreement with other studies(13,16,18,29).

The use of diagnostic imaging techniques for early diagnosis of MD in all patients with anorectal malformation will improve their treatment and prognosis, possibly increasing their chances of urinary and fecal continence and adequate neuromuscular development.

CONCLUSIONS

MRI is the exam of choice for the diagnosis of myelodysplasias. Myelodysplasia is found in association with high and low anorectal malformations. No direct relation was observed between myelodysplasia and the presence of heart, genitourinary and lower limb anomalies. The absence of associated vertebral malformation does not rule out the presence of MD. Urinary and fecal continence was not directly related with the presence of MD. Vertebral malformations, associated anomalies, urinary and fecal incontinence might indicate tethered cord.

REFERENCES