Variable expressivity of congenital longitudinal radial deficiency and spinal dysraphism in monozygotic twins

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We report monozygotic twins with longitudinal radial hypoplasia and low (L5) spina bifida occulta, but with differing severity. There is only one previous report of similar twins. We report these identical twins with the expression of longitudinal radial hypoplasia with mirror image pattern on contralateral sides, and the association of low spina bifida occulta.

Key words: monozygotic twins, radial agenesis, spina bifida, variable expression.

Structural abnormalities occur more commonly in monozygotic twins than in dizygotic twins and singletons. A common etiology for the monozygotic twinning and early embryonic malformations has been put forward by various authors as a possible explanation. Although many of the structural abnormalities in monozygotic twins are consequent to incomplete twinning, placental status or fetal constraint, the causative factor(s) that gave rise to monozygotic twinning may also lead to individual “early” malformation complexes. Sacrococcygeal teratoma, sirenomelia¹, VATER association, extrophy of the cloaca, holoprosencephaly, and anencephaly have been noted in monozygotic twins.

Goldenberg³ reported possible identical twin boys with bilateral complete absence of the radius and with spina bifida. To our knowledge there are no reports in the literature on longitudinal radial deficiency and spina bifida in monozygotic twins. We report for the first time variable expression of radial longitudinal deficiency and spinal dysraphism in monozygotic twins.

Case Report

These 20-month-old male twins were born to nonconsanguineous parents after a 33-week gestation to a 21-year-old primiparous mother by cesarean section due to transverse presentation. The placental status was not recorded. The birth weight of twin A was 2300 g and of twin B 2600 g. Physical examination revealed no abnormality of the head, thorax, back, hips, lower extremities, external genitalia or abdomen in either twin, and phenotypic appearance of both twins was identical (Fig. 1). The skin over the lumbosacral region was normal in both twins. There was no evidence of neural dysfunction in the lower extremities in either twin. The right forearm of twin A was 3 cm shorter than the left and his right thumb was hypoplastic, compatible with type 3B thumb hypoplasia⁴. His right elbow was stable and had normal range of motion. The left forearm of twin B was 1 cm shorter and radial styloid was more proximal than the ulnar styloid; his left hand was normal. Left elbow of twin B was stable and had normal range of motion (Fig. 2). Clinical findings of the twins are summarized in Table I.

Radiological examination of the forearm of both twins was compatible with type A hypoplasia of the radius, but the defect was milder in twin B (Figs. 3, 4). Radiological evaluation of the spine established the diagnosis of spina bifida occulta at L5 (Fig. 5); there were no significant spinal alignment abnormalities in sagittal and coronal planes in either twin. Spina bifida was more prominent on X-rays in twin A (Em) than in twin B (En). Abdominopelvic
Table I. Clinical Findings in Twins

<table>
<thead>
<tr>
<th></th>
<th>Birth weight</th>
<th>Weight at 20th month postnatal</th>
<th>Height at 20th month postnatal</th>
<th>Head circumference at 20th month postnatal</th>
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</thead>
<tbody>
<tr>
<td>Twin A (Em)</td>
<td>2600 g</td>
<td>11,800 g</td>
<td>85 cm</td>
<td>48 cm</td>
</tr>
<tr>
<td>Twin B (En)</td>
<td>2800 g</td>
<td>12,190 g</td>
<td>84 cm</td>
<td>48.5 cm</td>
</tr>
</tbody>
</table>

Fig. 1. Photograph depicting congenital radial deficiency more prominent in twin A (Em) (left) than in twin B (En) (right).

Fig. 2. Photographs of the forearms of the twins showed more severe involvement in twin A (Em) (left) than in twin B (En) (right).

Fig. 3. Roentgenograms of the right and left forearms of twin A (Em) (upper) and of twin B (En) (lower) demonstrate the relative shortness of the right forearm of twin A (Em).

Fig. 4. Roentgenograms of the right hand of twin A (Em) (left) and left hand of twin B (En) (right): note the delayed appearance of the distal ossification center of the right radius in twin A and left radius in twin B.

Fig. 5. Spina bifida (L5) demonstrated in both twins, more prominent in twin A (Em) (left) than in twin B (right).
ultrasonography and echocardiography revealed no abnormal findings in either twin. Both twins had XY karyotype. Blood group of both twins was A rh(+). HLA investigation showed that both twins had the same typing, HLA-ABC, A11, A23\textsuperscript{9}, B35, B49\textsuperscript{21}, CW7. Blood subgroups were identical, and are shown in Table II.

<table>
<thead>
<tr>
<th>Twin A (Em)</th>
<th>C+</th>
<th>Fy9-</th>
<th>JK9-</th>
<th>M+</th>
<th>P-</th>
</tr>
</thead>
<tbody>
<tr>
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<td>Fyb+</td>
<td>Jkb+</td>
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</table>

**Table II. Blood Subgroups of Twins**

Discussion

Early embryonic malformations such as caudal regression\textsuperscript{5}, VATER anomalies\textsuperscript{4} and diastematomyelia\textsuperscript{2} sirenomelia have been reported in monozygotic twins\textsuperscript{1} and siblings\textsuperscript{2}. Radial deficiencies are uncommon. We could find only one report of twins with congenital longitudinal radial deficiencies presenting with complete absence of the radius and associated spina bifida\textsuperscript{3}.

The twins presented in our paper are genetically monozygotic as shown by the blood grouping. We also collected DNA samples of the twins for future investigations. In our cases, the severity of congenital longitudinal radial deficiencies was different and manifested on opposite sides of the body. In twin A (Em), the metacarpus, phalanges and musculature of the right thumb were hypoplastic, while twin B (En) had only mild congenital longitudinal radial deficiency of the left side and his thumb was nearly normal in appearance.

Spina bifida was more severe in twin A (Em) than in twin B (En). A possible explanation is variable expressivity. We previously reported two sisters with variable expressivity in diastematomyelia\textsuperscript{2}.

In conclusion, we report for the first time genetic heterogeneity of congenital longitudinal radial deficiencies in monozygotic twins. Further similar observations will clarify the issue.

**REFERENCES**