Jarcho-Levin Syndrome with Diastematomyelia: Case Report of an Adult Patient and Review of Literature

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INTRODUCTION

Jarcho-Levin syndrome (JLS) is a rare congenital dysostosis characterized by multiple vertebral and costal anomalies. The combination of JLS and neural tube defect is rare. Only six cases of JLS accompanying diastematomyelia have been reported; all were in infants or children. We present the case of a 37-year-old female patient with JLS who also had diastematomyelia in lumbar vertebral level. This is the seventh case of JLS with diastematomyelia, and the first adult case.

Index terms
Jarcho-Levin Syndrome
Diastematomyelia
Congenital Dysostosis

We report the seventh case of JLS with diastematomyelia. It is the first case involving an adult patient.

CASE REPORT

A 37-year-old female visited our hospital for consultation for surgical treatment of thoracolumbar scoliosis. She had a short trunk and neck, with relatively long arms and a slightly protuberant abdomen. Whole spine scanogram showed thoracolumbar scoliosis and extensive malsegmentation of the thoracolumbar spines. Numerical and various intrinsic rib anomalies including irregular fusion and irregular narrowing were also noted on the left side (Fig. 1A).

Axial computed tomography of the spine using an Aquilion 64 (Toshiba Medical Systems, Tokyo, Japan) revealed the spina bifida at the level of the L3 vertebra (Fig. 1B) and diastematomyelia with a large intraspinal bony septum at the L4 vertebral level.
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(Fig. 1C). Volume rendering image reconstruction using an Aquarius iNuition ver 4.4.7 (Terarecon, Foster City, CA, USA) demonstrated a variety of vertebral deformities including hemi-vertebrae, block vertebrae, butterfly vertebrae and tripedicular vertebra at thoracolumbar spines (Fig. 1D). Abnormal fusion of the left ribs was also noted. The findings were compatible with spondylocostal dysostosis. There was evidence of diastematomyelia with a large intraspinal bony bar at the L4 level. Spinal magnetic resonance images demonstrated the split cord malformation at the whole thoracolumbar vertebral level. Axial T2-weighted scan at the level of T2 showed the separated spinal cord into two hemicords (Fig. 1E). Coronal T2-weighted image revealed a large intraspinal bony septum (Fig. 1F) with separation of the spinal cord into two hemicords.

Fig. 1. A 37-year-old female had Jarcho-Levin syndrome with diastematomyelia. 
A. Whole spine scanogram showed thoracolumbar scoliosis and extensive malsegmentation of the thoracolumbar spines. Numerical and various intrinsic rib anomalies including irregular fusion (arrows), irregular narrowing were also noted on the left side. 
B–D. Axial computed tomography (Aquilion 64, Toshiba Medical Systems, Tokyo, Japan) of spine demonstrated the spina bifida at the level of L3 vertebra (B, curved arrows) and diastematomyelia with a large intraspinal bony septum at L4 vertebral level (C, arrow). Volume rendering reconstruction (Aquarius iNuition ver 4.4.7, Terarecon, Inc.) image (D) demonstrated that variety of vertebral deformities including hemivertebrae (arrowheads), block vertebrae (asterisks) in thoracolumbar spines. And it also demonstrated multiple fusion of ribs on the left side (D, arrows). 
E, F. Spinal magnetic resonance images demonstrated the split cord malformation at the whole thoracolumbar vertebral level. Axial T2-weighted scan at the level of T2 shows the separated spinal cord into two hemicords (E, arrow). Coronal T2-weighted image reveals a large intraspinal bony septum (F, black arrow).
We report the seventh case of JLS with diastematomyelia. The patient is the oldest to date. Our case is significant, because the exact lifespans of the reported six cases of JLS with diastematomyelia are not known.

REFERENCES

척수이분증을 동반한 Jarcho-Levin 증후군: 성인 증례 보고와 문헌 고찰

김영선·이지혜·강미진·배경은·김재형·정명자·김성희·김지영·김수현·이한비

Jarcho–Levin 증후군(이하 JLS)은 척추와 늑골에 다양한 이상이 나타나는 드문 선천성 이골증이다. JLS와 신경관 결손이 동반된 경우는 드물며, 척수이분증이 동반된 JLS는 지금까지 여섯 증례만이 보고되었다. 또한, 이 여섯 증례들은 모두 유아나 소아 환자였다. 본 증례 보고에서는 37세 여성 환자의 증례를 보고하고자 한다. 이는 척수이분증이 동반된 JLS의 일곱 번째 증례이자 첫 번째 성인 환자 증례이다.

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