Incomplete cauda equina syndrome in adult monozygotic twins

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Abstract
Introduction Genetic factors play a dominant role in multifactorial etiology of disc degeneration as suggested in recent twin and familial aggregation studies that ultimately led to the identification of several associated gene forms. We report a case of adult monozygotic twin brothers with incomplete cauda equina syndrome due to central disc herniation of the lumbosacral junction.

Methods Twin No. 1 was admitted due to asymmetric bilateral sciatica, partial urinary retention and constipation, bilateral sensory deficits and rapidly progressing right foot paresis. Within two years twin No. 2 was admitted due to sudden bilateral sciatica, complete urinary retention, saddle and genital paresthesia and left calf hypesthesia. Preoperative radiologic work-up revealed a massive central L5-S1 herniated disc in both cases.

Results We performed an urgent L5-S1 discectomy on both twins. Six months after the procedure an elective L5-S1 TLIF with L4-S1 transpedicular screw fixation was performed on twin No. 1 due to L5-S1 reherniation and L5 right inferior articular process fracture. Both patients showed a complete neurologic recovery at follow-up visits.

Conclusion To our knowledge this is the first reported case of cauda equina syndrome in monozygotic twins of similar age, habits and occupational background thus supporting the evidence of genetic factors playing a primary role in symptomatic degenerative disc disease.
Case report

A 34 year old firefighter and nonsmoker with a history of two year duration of backache and left leg pain experienced worsening of pain in 2010 with bilateral sciatica. MRI showed an L5-S1 central and right recess herniated disc and he was admitted for elective surgery of disc evacuation (Picture 1).

![MRI of Twin No. 1 before L5-S1 discectomy](image)

**Picture 1: MRI of Twin No. 1 before L5-S1 discectomy**

During the first night in hospital he experienced worsening of pain in both legs, constipation and partial urinary retention. He had bilateral L5 and S1 dermatome hypesthesia (right leg more than left) with progressive right foot toe dorsiflexion paresis and right foot plantar flexion paresis. We diagnosed the condition as partial cauda equina syndrome with urinary retention and an urgent L5-S1 discectomy was performed. The patient made a gradual early postoperative neurologic improvement but one month after the procedure, due to an early return to work, his backache and right leg pain returned. Upon examination he had a right foot dorsiflexion weakness (3/5) and left foot hypesthesia that improved with physiotherapy. Upon follow up visits he complained of persisting bilateral sciatica and CT scan of lumbosacral spine revealed right L5 inferior articular process fracture, L5-S1 central and posterolateral rehemiation and L4-L5 lateral canal stenosis.

An elective L4-S1 decompression and fusion was performed and at follow up visits a complete neurologic recovery was noted but his rehabilitation was delayed due to chronic pain with remissions (Picture 2).

Two and a half years later his identical twin brother, a construction worker and nonsmoker with five year history of bilateral sciatica experienced a sudden lower back and left leg pain with five hours of complete urinary retention. He also complained of saddle, genital and right leg paresthesia. Upon examination he had a positive 20° left and 30° right straight leg rise test, left calf hypesthesia and perineal dysesthesia and during urinary catheterization more than 1000 ml of urine retention was noted. An urgent CT scan of lumbosacral spine revealed a central L5-S1 herniated disc (Picture 3).
Again we diagnosed the condition as partial cauda equina syndrome with urinary retention and an urgent L5-S1 discectomy was performed. After surgery we noted an excellent neurologic recovery and the patient remained pain free at follow up visits.

Picture 2: X-ray of Twin No. 1 after L4-S1 decompression and fusion

Picture 3: CT of Twin No. 2 before discectomy; notice the similarities in radiographic changes between Twin No. 1 and Twin No. 2
Discussion

The PubMed and Cochrane library database search found five case reports of disc herniations in monozygotic twins (Table). All monozygotic twins in those case reports had the same segment disc herniations with almost synchronous occurrence of symptoms and almost identical radiographic findings on other lumbar levels indicating strong genetic factors at play.

<table>
<thead>
<tr>
<th>Author/published</th>
<th>Age of onset</th>
<th>Sex</th>
<th>Timing between herniations</th>
<th>Segment</th>
<th>Herniated side</th>
<th>Radiographic/operative findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gunzburg et al/1990</td>
<td>13</td>
<td>Female</td>
<td>Few months</td>
<td>L5-S1</td>
<td>Different</td>
<td>1 extrusion, 1 protrusion</td>
</tr>
<tr>
<td>Matsui et al/1990</td>
<td>16/18</td>
<td>Female</td>
<td>2 years</td>
<td>L4-L5</td>
<td>Different</td>
<td>1 protrusion, 1 extrusion</td>
</tr>
<tr>
<td>Bhardwaj et al/2004</td>
<td>52</td>
<td>Female</td>
<td>4 months</td>
<td>L5-S1</td>
<td>Same</td>
<td>Both extrusions</td>
</tr>
<tr>
<td>Dhir et al/2009</td>
<td>41</td>
<td>Female</td>
<td>Few months</td>
<td>L4-L5</td>
<td>Same</td>
<td>1 extrusion, 1 protrusion</td>
</tr>
<tr>
<td>Nemoto et al/2012</td>
<td>24</td>
<td>Male</td>
<td>3 months</td>
<td>L4-L5</td>
<td>Same</td>
<td>Both protrusions</td>
</tr>
</tbody>
</table>

The basis of research on the role of genetics in degenerative disc disease in the last twenty years were twin studies that focused on relationships between environmental risk factors and T2 weighted MRI changes of loss of disc signal intensity, loss of disc height and disc bulging. According to the results of a Finnish Twin Cohort study on 115 matched monozygotic male twin pairs that were discordant for environmental factors, familial aggregation accounts for 77% of variability in degenerative disc disease scores for upper lumbar spine and 43% of the same variability for lower lumbar spine (Battié, Videman, Gibbons et al). Comparing 86 monozygotic and 77 dizygotic twins, Sambrook et al reported a 74% genetic contribution for lumbar degenerative disc disease. According to preliminary results of Twin Spine Study on 147 monozygotic and 153 dizygotic twin pairs, heritability estimates for degenerative disc disease are close to 50% (Battié, Videman, Kaprio et al). Genetics clearly plays a dominant role in disc degeneration with stronger effects being associated with an earlier onset (Zhang et al).

Based on results of twin studies the researchers in the last 20 years focused on discoveries of allelic gene forms associated with degenerative disc disease. The four major categories of genes related to lumbar disc disease are: structural components of the intervertebral disc (collagen type I, III, IX, XI, aggrecan), degradation enzymes of the disc matrix (matrix metalloproteinase, interleukins), genes related to bone structure (vitamin D receptor, estrogen receptor) and other genes such as Sox9 that regulates the expression of COL2A1, aggrecan and collagen IX genes.

Conclusion

Lumbar degenerative disc disease is according to twin and familial aggregation studies a multifactorial and polygenic condition with genetic factors playing a dominant role in its manifestation. Disc herniations in monozygotic twins are according to sporadic case reports located in the same segment of the lumbar spine and are time dependent, occurring almost synchronously. To our knowledge this is the first reported case of
incomplete cauda equina syndrome in monozygotic twins due to lumbar disc herniation.

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**Literature**


