Currarino Syndrome and Pediatric Presacral Tumors

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Currarino Syndrome (CS) Overview

- First described by Dr. Guido Currarino in 1981
- Form of caudal regression syndrome
- Triad with associated anomalies
  - Anorectal malformation
  - Sacrococcygeal osseous defect
  - Presacral mass
- Chronic constipation
  - Often first and only sign and may persist post-operatively
CS Overview

- **Classification**
  - Complete: full expressivity with early presentation
  - Mild: hemisacrum + one other anomaly
  - Minimal: hemisacrum only

- **Epidemiology**
  - 2:1 female predominance in children
  - 80% of patients diagnosed before age 12
  - Malignancy reported in six of 300 patients with diagnosis
  - Limited long-term data
Genetic Basis of CS

- HLXB9 homeobox gene at 7q36
  - Point mutation or entire sequence deletion
  - Autosomal dominant inheritance
  - 50% of patients have a family member with 1+ components

- Phenotypic variability
  - Hemisacrum → classic triad → other anomalies
Anorectal Malformation

- Rectoperineal fistula or anorectal stenosis
  - Most common in CS
- Rectourethral fistula
- Rectovestibular fistula
- Rectocloacal fistula
Sacral Agenesis

- Type I - Total sacral agenesis
- Type II - Total sacral agenesis without lumbar vertebral involvement
- Type III - Subtotal sacral agenesis (S1 present)
- Type IV - Hemisacrum
  - “Scimitar sacrum”
- Type V - Coccygeal agenesis only

Adapted from Martucciello et al., J Pediatr Surg 2004
Presacral Mass

- Boundaries of presacral space
- Embryologic significance of contents
- Presentation is dependent on mass location

Adapted from Aranda-Narváez et al., WJGS 2012
Sacrococcygeal Germ Cell Tumors

- Altman Classification
  - Type I - Mostly external
    - 50% cases
  - Type II - External with significant intrapelvic component
  - Type III - External with pelvic and abdominal components
  - Type IV - Intra-pelvic and intra-abdominal
  - 80% diagnosed by six months

Adapted from Kocaoglu et al., Radiographics 2006
Sacrococcygeal Germ Cell Tumors

- Imaging characteristics
  - Mature, cystic → benign
  - Immature, solid → malignant

- Serum evaluation
  - Alpha fetoprotein elevated in 50% malignant teratomas
  - Beta human chorionic gonadotropin elevation in choriocarcinomas

Adapted from Martucciello et al., J Pediatr Surg 2004
Anterior sacral meningocele

- Due to dural herniation through sacral foramen/defect
- If symptomatic, then related to mass effect, neurologic compromise, meningitis, or rupture
- Require pre-operative imaging of any nerve roots within hernia sac
Other Categories of Masses

- Dermoid cyst
- Enteric cysts
- Cystic lymphatic malformation
- Ganglion cell or neuroblastic tumors
- Pelvic abscesses
- Sarcomas and other mesenchymal masses
- Primary sacral tumors with presacral extension
Surgical Approaches

- Wide, *en bloc* resection of presacral tumor and coccyx
- Anterior intra-abdominal approach
  - Lowest tumor extent above S4 without nerve involvement
- Posterior midsagittal perineal approach
  - Type I and II tumors
- Combined posterior and intra-abdominal approach
  - Type III and IV tumors

### Table 2: Large series of presacral tumors

<table>
<thead>
<tr>
<th>Author (yr)</th>
<th>Patients</th>
<th>Period (yr)</th>
<th>Male/female</th>
<th>Age (yr)</th>
<th>Benign/malignant</th>
<th>Surgical approach (A/P/C)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Uhlig <em>et al</em> 1975</td>
<td>63</td>
<td>30</td>
<td>17:46</td>
<td>-</td>
<td>37/26</td>
<td></td>
</tr>
<tr>
<td>Jao <em>et al</em> 1985</td>
<td>120</td>
<td>20</td>
<td>46:74</td>
<td>43 (0-81)</td>
<td>69/51</td>
<td>21/79/2 (85% resectibility rate)</td>
</tr>
<tr>
<td>Wang <em>et al</em> 1995</td>
<td>45</td>
<td>15</td>
<td>20:25</td>
<td>41 (15-76)</td>
<td>23/22</td>
<td>24/13/6 (95% resectibility rate)</td>
</tr>
<tr>
<td>Lev-Chelouche <em>et al</em> 2003</td>
<td>42</td>
<td>10</td>
<td>14:28</td>
<td>40 (21-84)</td>
<td>21/21</td>
<td>18/21/3</td>
</tr>
</tbody>
</table>

A: Anterior; P: Posterior; C: Combined.

*Adapted from WJGS 2012 Aranda-Narváez*
Diagnostic Algorithm

- History and physical
  - Anorectal malformation
  - Severe, unexplained constipation

- XR sacrum, two views
  - Hemisacrum is pathognomonic

Sacral Ratio Index
Adapted from <http://www.springerimages.com/Images/MedicineAndPublicHealth/1-10.1007_s11751-011-0109-0-1>
Diagnostic Algorithm

- If hemisacrum is present:
  - HLXB9 analysis
  - XR sacrum of family members
    - Sporadic vs. familial
- Cardiac evaluation
- U/S of urinary tract
- Rectal suction biopsy
  - Dysganglionoses, including Hirschsprung’s
- Pelvic/spinal MRI
  - Operative planning
  - CS classification
  - Identify neural tube defects
    - Tethered cord
- Utility of prenatal U/S or fetal MRI
Treatment Algorithm by CS Type

Adapted from Martucciello et al., J Pediatr Surg 2004
Case Example - Patient I.S.

- Female born with a sacral dimple and cutaneous hemangioma
  - MRI at six and nine months demonstrating pre-sacral mass
  - Abnormal facies attributed to question of trisomy 1

- Outpatient visit at 10 months
  - 1.9 x 2.6 x 2.5cm presacral lipomatous mass
  - Asymptomatic anterior displacement of rectum and anus
  - Tethered cord
  - Non-compliance in follow-up

- Phone call at 17 months
  - Constipation
Case Example - Patient I.S.

- ED presentation at 23 months
  - Antalgic gait
  - Change in stool character and consistency
  - Concern for lower back pain

- Contrast MRI concerning for 5.0 x 4.9 x 4.6cm presacral heterogeneous mass and iliac lymph node enhancement

- Alpha feto-protein markedly elevated

- Tumor resection complicated by surgical site infection

- Staging CT demonstrative of left lung nodules and axillary/inguinal lymph node involvement and incidental bilateral SVC

- Diagnosis?
Conclusion

- Currarino syndrome is a heterogeneous set of HLXB9-associated anomalies with emphasis on sacral agenesis, presacral mass, and anorectal malformation
- Diagnosis requires clinical suspicion as well as both bony and soft tissue imaging
- Treatment is primarily operative but may require coordination with multiple surgical subspecialties
References


