6 Urinary Problems Associated with Imperforate Anus

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6.1 Introduction

The term "imperforate anus" includes all kinds of anorectal malformations from covering of the anus by a thin skin membrane to high anorectal atresia – with or without a fistula, into the urethra or bladder – and to cloacal anomalies in females. It is well known that a high proportion of patients with imperforate anus have this maldevelopment in association with anomalies of one or several other organ systems. Among those, urinary tract anomalies with or without infections, as well as functional disorders of the urinary system, are common and may cause serious complications. Thus, the overall morbidity and mortality of patients with anorectal malformations is considerably influenced by the associated urinary tract anomalies.

To facilitate the understanding of the problem, we briefly describe the different types of imperforate anus and the incidence of associated anomalies before we focus on the related urinary tract problems.

6.2 Anorectal Malformations

In earlier years anorectal malformations were classified into two subtypes: a high and low form, depending on whether the distal rectal pouch ended above or below the levator muscle level. On the occasion of the international Wingspread workshop meeting in 1984, the different types of imperforate anus were classified into three major groups and into the male and female patterns of the malformation (Stephens and Smith 1986). Briefly, a high–intermediate–low classification was agreed upon, and minor and rare subtypes were omitted. “High” anomalies are characterized by anorectal agenesis or rectal atresia with or without a rectovesical or rectoprostatic fistula in males or a rectovaginal fistula in females. The blind rectal pouch ends definitely above a hypotrophic puborectalis muscle sling.

In “intermediate” malformations, the rectal pouch enters that sling; there may be a rectobulbar fistula in males and a rectovestibular or rectovaginal fistula in females. In the “low” forms the rectum passes through a well-developed puborectalis muscle and may end in an anocutaneous fistula in males and an anovestibular or anocutaneous fistula in females. The consensus conference placed the female cloaca in a separate group because it may be high, intermediate, or low depending on the length of the common channel. With regard to anal function and continence, it is evident that the high forms of imperforate anus have clearly less satisfying results than the low forms. Beyond that, the high and intermediate forms have a higher incidence of associated malformations and urinary tract functional disorders.

6.2.1 Embryology of Imperforate Anus

In the 4-week-old embryo, the hindgut expands to form the internal cloaca, into which issue the large intestine, the allantois, and the wolfian or
mesonephric ducts. The internal cloaca is separated from the external cloaca by the cloacal membrane. The partitioning of the internal cloaca by a cranio-caudally growing septum begins at the 4-mm stage and is completed at the 16-mm stage, when the septum reaches the cloacal membrane. Once the septum is completed, the cloaca is divided into a ventral urogenital sinus and the dorsal rectum. The Wolffian ducts become organized into the vasa deferentia and the vesicae seminales in the male, while in the female they are the leading structures for the proceeding of the müllerian ducts into the vestibule. The external cloaca is a depression of tissue formed by the bilateral genital folds and the genital tubercle on the ventral aspect. When the septum reaches the cloacal membrane, the latter atrophies and both systems enter the common external cloaca. The process of partitioning now extends caudally by the uro-anal septum. The high and intermediate groups of anorectal malformation can be seen as the result of a disturbed development of the partitioning of the internal cloaca with the gut ending in a fistula to the verumontanum or higher in males, and into the vagina or fossa navicularis of the vestibule in females. The low forms refer to developmental errors affecting the partitioning of the external cloaca, resulting in a fistula to the perineum or to the female vestibulum or a completely or partially persisting anal membrane (Stephens and Smith 1971).

Duhamel (1961) reported that the most frequent malformations associated with imperforate anus are just those which one finds constantly in the sirenomelia anomaly, and he concluded that the whole pattern of anorectal malformations belongs to the syndrome of caudal regression. This hypothesis is in agreement with the work of Berdon et al. (1966) and Elliot et al. (1970), who explained the common association of lumbosacral vertebral anomalies and hindgut malformations by a disturbed development of the notochordal organizer at a very early stage of embryogenesis (Fig. 6.1).

6.2.2 Associated Malformations

As mentioned above, there is agreement in the literature that anorectal malformations are highly associated with other anomalies of viscera or the skeletal system. The overall reported incidence varies from 20% to 70%, a range which depends largely on a careful and systematic search for additional anomalies (Stephens and Smith 1971). In a series of 75 patients we found an overall incidence of 72% further anomalies, reaching nearly 100% in the subgroup of deceased patients (Höllwarth and Menardi 1983) (Table 6.1). These findings confirmed the conclusion from a necropsy study of babies dying with anorectal anomalies (Moore and Lawrence 1952) that there is a nearly 100% association with other malformations.

Table 6.1. Associated malformations in 75 patients with anorectal malformations

<table>
<thead>
<tr>
<th>Anatomical system</th>
<th>Total (n=75) (%)</th>
<th>Survivors (n=59) (%)</th>
<th>Deceased (n=16) (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Urinary tract</td>
<td>41.3</td>
<td>35.5</td>
<td>62.5</td>
</tr>
<tr>
<td>Genitalia</td>
<td>12.0</td>
<td>10.1</td>
<td>18.7</td>
</tr>
<tr>
<td>Skeletal system</td>
<td>46.6</td>
<td>37.2</td>
<td>37.5</td>
</tr>
<tr>
<td>Cardiac system</td>
<td>18.6</td>
<td>11.8</td>
<td>37.5</td>
</tr>
<tr>
<td>Intestinal tract</td>
<td>18.6</td>
<td>8.4</td>
<td>62.5</td>
</tr>
<tr>
<td>Cerebral</td>
<td>13.3</td>
<td>8.4</td>
<td>25.0</td>
</tr>
<tr>
<td>Others</td>
<td>22.6</td>
<td>20.3</td>
<td>31.2</td>
</tr>
<tr>
<td>Total</td>
<td>72.0</td>
<td>65.5</td>
<td>100</td>
</tr>
</tbody>
</table>

Very frequently more than one organ system is involved. This is also reflected by the so-called VATER or VACTERL association that combines vertebral anomalies, anorectal atresia, cardiac failure, tracheoesophageal fistula, esophageal atresia, renal dysplasia, and limb malformation.