remarkably from only published
The stool between condition.
R. V. aetiology of The
found to have stool water 'constipation'.
Stool water stenosis (PS) is largely unknown. It has long been
but the man's parents suggested that the man's parents
none of his
A genealogical table of infantile hypertrophic pyloric
infantile hypertrophic pyloric
3
and decrease
as expected there was a wide range of individual variation in stool weight and frequency, but the general trend of an increase in weight and decrease in frequency with age is apparent. However, even in the second year it was common for the children to pass more than one stool a day, although some children went for 2–3 days without defecation in the absence of clinical 'constipation'. Stool water content hardly varied from the first week to the second year and had a remarkably small SD. There was no correlation between stool frequency and stool weight.

Comment
The only published information on defecation patterns

<table>
<thead>
<tr>
<th>Table</th>
<th>Data on stool frequency, weight, and water content in normal infants at different ages</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1st week</td>
</tr>
<tr>
<td>No. of infants</td>
<td>16</td>
</tr>
<tr>
<td>No. of stools</td>
<td>278</td>
</tr>
<tr>
<td>Stool frequency</td>
<td>Mean interval between stools (hours) ± 1 SD</td>
</tr>
<tr>
<td>Mean</td>
<td>5.2 ± 1.9</td>
</tr>
<tr>
<td>Range</td>
<td>9.9 ± 6.5</td>
</tr>
<tr>
<td>±1 SD</td>
<td>13.2 ± 9.2</td>
</tr>
<tr>
<td>0.5–22</td>
<td>14.9 ± 8.3</td>
</tr>
</tbody>
</table>

641 stools were examined. As expected there was a wide range of individual variation in stool weight and frequency, but the general trend of an increase in weight and decrease in frequency with age is apparent. However, even in the second year it was common for the children to pass more than one stool a day, although some children went for 2–3 days without defecation in the absence of clinical 'constipation'. Stool water content hardly varied from the first week to the second year and had a remarkably small SD. There was no correlation between stool frequency and stool weight.

Infantile hypertrophic pyloric stenosis—unusual familial incidence

V. R. FINSEN
Department of Surgery, Paediatric Section, Regionyskehuset, Trondheim, Norway

SUMMARY A man, who had been treated for infantile hypertrophic pyloric stenosis (PS), was found to have 3 sons with the same condition. A genealogical table of his family showed that both his parents had relatives with PS. So far as it was known, none of his wife's relatives had the condition. It is suggested that the man's parents carried genes predisposing to PS and transmitted these to him in such quantity that he and all his children acquired the condition.

The aetiology of infantile hypertrophic pyloric stenosis (PS) is largely unknown. It has long been recognised that there is an increased preponderance of the condition in the relatives of patients. This familial predisposition is shown very clearly in the family to be described.

A patient (IV₁ in the Figure) had an operation at this hospital some years ago for PS. On further investigation it was found that both his brothers (IV₂ and IV₃) and his father (III₁) had been treated for the same condition. The main features of their case histories are summarised in the Table.

A genealogical table stretching over four generations was assembled for this family (Figure). The patient's mother had no known relatives with PS, but both his paternal grandfather and grandmother (II₃

References

Correspondence to Dr O. G. Brooke, Department of Child Health, St George's Hospital, Cranmer Terrace, London SW17 0RE.
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Table Main features of the case histories of Case IIIb and his 3 sons IV3, IV4, and IV5

<table>
<thead>
<tr>
<th>Case</th>
<th>Gestation at birth (weeks)</th>
<th>Start of projectile vomiting</th>
<th>Clinical findings</th>
<th>Treatment</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>IIIb</td>
<td>40</td>
<td>3rd week</td>
<td>Case notes lost</td>
<td>Spasmalytics and frequent small feeds</td>
<td>Projectile vomiting ceased after few weeks. Uneventful recovery</td>
</tr>
<tr>
<td>IV3</td>
<td>40</td>
<td>2nd day</td>
<td>Tumour palpated. Hypertrophy on x-ray</td>
<td>Pyloromyotomy at 4 days</td>
<td>Reoperated next day because of gastric retention. Diaphragm in pyloric region excised. Uneventful recovery</td>
</tr>
<tr>
<td>IV4</td>
<td>38</td>
<td>3rd week</td>
<td>Tumour palpated. Visible peristalsis. X-ray normal</td>
<td>Pyloromyotomy at 3 weeks</td>
<td>Uneventful recovery</td>
</tr>
<tr>
<td>IV5</td>
<td>40</td>
<td>3rd day</td>
<td>First x-ray normal, Hypertrophy on x-ray after 10 days</td>
<td>Pyloromyotomy at 2 weeks</td>
<td>Uneventful recovery</td>
</tr>
</tbody>
</table>

and IIb) had. Cases IIIb and IV3 had both had pyloromyotomies performed, and IIIb had died on his way to hospital when he was 7 weeks, after suffering projectile vomiting and progressive weight loss from age 2 weeks.

There was no history of marriage between relatives.

Discussion

Although PS is found in infants it is not congenital in the sense that it is present at birth. The symptoms usually present between ages 3 and 6 weeks, and only rarely during the first week. All reported series show a pronounced preponderance of male to female patients, and it has long been noted that there is an increased incidence of PS in the relatives of patients who have the condition. Carter and Evans (1969) studied 813 patients treated by Ramstedt’s pyloromyotomy between 1920 and 1949; 20% of the sons and 7% of the daughters of their female patients had developed PS, but only 5% of the sons and 2.5% of the daughters of the male patients had developed the condition. They estimated the incidence in the general population as being 0.5% for liveborn boys and 0.1% for liveborn girls. They concluded that the hereditary background to PS is polygenic.

It would appear that there are a number of genes in different locations which may predispose for PS, and that it is necessary for a certain number of these genes to code for the condition for it to develop. The female phenotypes seem to be protected to some degree so that a greater genetic load is required for girls to acquire the disease. This greater genetic load explains why the children of female patients are more likely to develop PS rather than those of male patients.

It is interesting to note that Cases IV3 and IV5, who were born at term, developed the symptoms at an exceptionally early age. There was little time for any postnatal environmental factors to influence the development of the condition in them. Their brother, Case IV4, was born 2 weeks preterm and PS was diagnosed at age 3 weeks. All 3 brothers, therefore, developed the condition at about the same time after conception.

It is not certain from whom Cases IIIb, IIIb, and IV3 acquired their predisposition for PS. However, finding the condition in these patients increases the likelihood of Cases IIIb and IIIb carrying genes predisposing to PS, although not to such a degree that they developed the condition. These genes were inherited by their son (Case IIIb) in such quantity, that not only did he develop PS, but he transferred the condition to all his 3 children.

Reference


Correspondence to Dr V. R. Finsen, Dalen Hageby 8, 7000 Trondheim, Norway.
Infantile hypertrophic pyloric stenosis--unusual familial incidence.

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