Diagnosis of Hirschsprung’s disease by punch biopsy of rectum

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In this paper we present evidence that punch biopsy of the rectum is a simple, safe, and reliable method of diagnosing Hirschsprung’s disease.

Patients and methods
Between January 1970 and December 1974 a total of 234 punch biopsies of the rectum and colon were performed on 112 children. In 102 of the children the biopsy was to try to establish a diagnosis, whereas in the remaining 10 Hirschsprung’s disease had already been diagnosed and serial biopsies were done to try to define the aganglionic segment. The age distribution of the children at the time of biopsy is shown in the Table.

<table>
<thead>
<tr>
<th>Age</th>
<th>No. of patients</th>
<th>Hirschsprung’s disease diagnosed</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;1 week</td>
<td>19</td>
<td>5</td>
</tr>
<tr>
<td>1-3 weeks</td>
<td>18</td>
<td>7</td>
</tr>
<tr>
<td>1-11 months</td>
<td>38</td>
<td>15</td>
</tr>
<tr>
<td>1-3 years</td>
<td>24</td>
<td>5</td>
</tr>
<tr>
<td>4-12 years</td>
<td>13</td>
<td>3</td>
</tr>
<tr>
<td>Total</td>
<td>112</td>
<td>35</td>
</tr>
</tbody>
</table>

The technique used for taking rectal tissue samples was a modification of that described by Shandling (1961). Preparation of the bowel is unnecessary but suction through a 10 FG catheter helps to clean any liquid meconium or faeces from the operative site. Preoperative vitamin K is essential in neonates. The lithotomy position is most satisfactory in neonates while the left lateral position is more suitable in older children. A standard Chevalier-Jackson vocal cord biopsy forceps is used (Fig.). A view of the rectum is obtained in newborn infants by using a nasal speculum and in older patients by using a bivalve rectal speculum. A sigmoidoscope is used for serial biopsies higher up or via a colostomy.

With the forceps in position a bite of mucosa and submucosa enough to fill the cup is taken under direct vision. This action alone will not cut the specimen completely. While keeping the jaws firmly closed the instrument is sharply withdrawn. Specimens 3 to 5 mm in diameter are obtained in this way. Depending on the age of the patient, the samples for diagnostic biopsy are taken 2 to 3 cm above the dentate line to avoid the normal hypoganglionic zone (Aldridge and Campbell, 1968). It is advisable to take two specimens from each patient in case one of them contains insufficient submucosa. After this procedure bleeding is minimal and ceases spontaneously.

Anaesthesia. Biopsy was performed under general anaesthesia in 47 (42%) of the 112 patients. In 17 of the 47 the sample was taken at laparotomy or some other operative procedure for which the child was already

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patients grossly mentally retarded. One sprung's included that nerve appearing sudomotor cells. The epithelium was showed by colonic specimens within overnight. In the fourth patient a large pelvic haematoma developed resulting in urinary retention: this was due to a combination of the biopsy procedure coinciding with the onset of haemorrhagic disease of the newborn and pre-operative vitamin K having been accidentally omitted.

Discussion

Normally ganglion cells are present in both the submucous and myentericplexuses throughout the length of the bowel wall. At the anal end there is a hypoganglionic zone where ganglion cells may normally be scanty or absent (Bodian, Stephens, and Ward, 1949). In the myenteric plexus this zone extends proximally from the anal valves for, on average, a distance of 4 mm (0–14 mm). In the submucous plexus the corresponding distance is 10 mm (3–17 mm) (Aldridge and Campbell, 1968). In Hirschsprung's disease ganglion cells are absent from both plexuses in the affected segment of bowel.

Full thickness biopsy of the rectum, as described by Swenson, Fisher, and MacMahon (1965), is a technically difficult procedure and not without hazards, especially in neonates. General anaesthesia is always necessary. Bodian (1960) showed that full thickness biopsy was not necessary for the diagnosis of Hirschsprung's disease and that histological examination of the submucosal plexus only was sufficient. He recommended taking a specimen of mucosa and submucosa 0·5 cm × 2 cm. Several papers since have shown that much smaller biopsy specimens are perfectly adequate (Shandling, 1961; Dobbins and Bill, 1965; Campbell and Noblett, 1969) and Noblett (1969) devised a special suction instrument for obtaining specimens of the rectal mucosa.

A word of caution about the histological assess-
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