Hypertrophic Pyloric Stenosis: Adult and Congenital Types Occurring in the Same Family

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Both congenital and adult types of hypertrophic stenosis of the pylorus are recognized. The congenital variety has an incidence of 5 per 1000 male births and 1 per 1000 female births in the general population of England (Carter, 1967), but it is less frequent in Negroes and in Asians (Donovan and Stanley-Brown, 1962; Swan, 1961). Both environmental and genetic factors seem to operate in the aetiology of the disease (Carter, 1961).

The adult type is complex (Berk, 1963). The incidence of the disease is difficult to assess as there is variability in the awareness of the disorder, difference in the diagnostic criteria, and often misclassification when found in association with other anomalies. There are several views on the aetiology of adult hypertrophic pyloric stenosis. Some authors (Horwitz, Alvarez, and Ascanio, 1929; Raffensberger, 1955) have considered the possibility of it being secondary to gastritis, ulcer, cancer, or other gastric anomalies. Others (Atkinson et al., 1957) have suggested that prolonged spasms of the pylorus could lead to thickening of the pyloric muscle, but there is little evidence to support this. It is also held that the primary adult form of hypertrophic pyloric stenosis is the same condition as that of the congenital one, but milder in severity and of later expression (MacCann and Dean, 1950; North and Johnson, 1950; Keynes, 1965; McConnell, 1966)—a view supported by the presence of reports of cases of adult and of congenital hypertrophic pyloric stenosis occurring in one and the same family.

**Case Reports**

A 38-year-old woman (III.5 in Fig. 1) was first seen in August 1965. She had had for several years occasional epigastric discomfort and heaviness after meals, which was relieved by vomiting. A barium meal examination showed elongation of the stomach, with a large and narrow pyloric canal (Fig. 2) and a moderate delay in gastric emptying. The symptomatology reappeared early in 1967, progressively increasing in frequency and severity in mid-1967. The physical examination revealed a thin woman weighing 38 kg. (the previous year her weight had been 44 kg.). No tumour was palpable in the abdomen and a second barium meal study confirmed pyloric stenosis. A subtotal gastrectomy was performed, the patient recovered uneventfully and, since then, she has been free of symptoms. Pathological examination of the excised specimen showed thickening of the muscle fibres of the pylorus up to 1.7 cm. (Fig. 3). Microscopically it was judged as pyloric hypertrophy with muscular hyperplasia (Fig. 4).

The pedigree is shown in Fig. 1. The father of the propositus (II.3) had had a clinical history similar to his daughter. He was operated on in 1962 for a perforated peptic ulcer and died after operation. The specimen showed a perforated pre-pyloric ulcer and hypertrophic pyloric stenosis. A first cousin (III.9) and a daughter (IV.4) of the index case had required Ramstedt's operation during the first month of life. Another first cousin (III.1) had had a long history of vomiting but a barium meal study failed to reveal any abnormality. The propositus' brother (III.4) and another first cousin (III.8) were treated medically for

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After Carter’s studies (1961, 1965) the accepted mode of inheritance of the congenital type is that of multifactorial inheritance. He found that first degree relatives of male index patients were affected.

Discussion

There are only two previous published reports of both adult and congenital pyloric stenosis, occurring in the same family. Fenwick (1953) described a family in which a man with the adult form of the disease had two sons who were operated on early in infancy for the congenital type; he also had a nephew with congenital pyloric stenosis. Wong-Ming (1961) reported a family in which the father had adult pyloric stenosis and his second son had the congenital variety.

In our family there are two confirmed cases with the adult form, one (II.3) probably with the 'complicated type' of adult pyloric hypertrophy, and the other (III.5) with the 'primary variety'. There are, in addition, two proven cases with congenital pyloric stenosis (III.9 and IV.4). Two further cases were suspected of having a mild form of the congenital condition (III.4 and III.8), but unfortunately no x-ray studies were carried out; a long follow-up of them is obviously needed.

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Summary

The presence of two patients with congenital pyloric stenosis and two with the adult variety in the same family is described. This supports the view that both types have the same aetiology, and fits with the concept that the genetic predisposition to the condition is multifactorial.

References


Fig. 4. Microscopical view of the excised specimen showing pyloric hypertrophy, with muscular hyperplasia.

in a proportion of 4.6% for males, which is 10 times higher than the prevalence in the general population, and 2.3% for females (25 times higher). In relatives of female index patients there are 15-4% affected males (30 times higher) and 9-8% females (100 times higher). Twin studies have shown a higher rate of concordance in monozygotic than in dizygotic twins, though there is discordance in 30-50% of the monozygotic pairs (Powell and Carter, 1951; Metrakos, 1953; MacMahon and McKewon, 1955). These findings are all in accord with those expected in multifactorial genetic inheritance.

If we assume that both conditions have a similar aetiology, it can be argued that patients with just enough polygenic load to reach the threshold for pyloric hypertrophy would not have symptoms in infancy but would develop the disease later on. This hypothesis would also explain the rarely seen positive family history in affected adults, as these patients would have fewer genes for the disease (McConnell, 1966), and their relatives would not share enough of them to develop pyloric hypertrophy.
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