

Apple Peel Syndrome (Congenital Intestinal Atresia)

A Family Study of Seven Index Patients

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The type of jejunal atresia with agenesis of the mesentery, which has been variously described as the 'Christmas tree', 'maypole', or 'apple peel' deformity, consists of a duodenal or high jejunal atresia associated with absence of the small bowel mesentery and pre-arterial arcades of the superior mesenteric artery. The resulting appearance is of distal small bowel coming straight off the caecum and twisted around a marginal artery (Fig.), suggesting to different observers the designations quoted. The affection has been described in isolated cases (Santulli and Blanc, 1961; Jiminez and Reiner, 1961; Weitzman and Vanderhoof, 1966) but not, as far as the authors know, in more than one member of a sibship. The present paper records a family study of 7 index patients with this rare malformation, belonging to 6 families.

Material and Methods

The index patients, none of whom survived, were children admitted to The Hospital for Sick Children, Great Ormond Street, London, or to Paddington Green Children's Hospital, London, between 1949 and 1967. These children are believed to comprise the total of such cases admitted to these hospitals for surgery during this period, and they therefore form an unselected and consecutive series. A search of the post-mortem records for the two hospitals from 1968 back to 1948 failed to reveal any other similar cases. In fact there was a total of 8 index patients in 7 families, but in 1 instance, the affected child proved to be the second illegitimate baby of a young mother who had deliberately concealed the baby's birth from her parents. It was decided not to attempt to contact this mother lest such inquiries should cause undue anxiety.

The parents in each of the remaining 6 families were visited at their homes, and, whenever possible, the brothers and sisters of the affected children were seen.

Results

Sibship data are set out in the Table. Of the 7 index patients in 6 sibships, 3 were male and 4 were female. In addition to the sibship (Family 1) in which both affected children were index patients, one other sibship (Family 3) contained two affected children. In this family an older sib of the index patient had died at another hospital following operation on the 10th day of life, and necropsy only confirmed the diagnosis already made at operation.

There was no instance of parental consanguinity.

Chromosome studies carried out on the parents in Family 1 showed a normal karyotype in each case.



FIG. Typical 'apple peel' appearance of small bowel at operation.

TABLE
SUMMARY OF SIBSHIPS

| Family No. | Details of Sibships | Total Affected Sibs | | Total Unaffected Sibs | | Total Sibs | | Parents' Date of Birth | |
|------------------------|---|---------------------|---|-----------------------|---|------------|---|------------------------|--------|
| | | M | F | M | F | M | F | Mother | Father |
| 1 | <i>F*</i> 10.63 (d 12 days); <i>F*</i> 5.65 (d 4 days) | - | 2 | - | - | - | 2 | 11.37 | 10.37 |
| 2 | <i>F*</i> 7.66 (d 25 days); M 6.67 | - | 1 | 1 | - | 1 | 1 | 7.44 | 11.45 |
| 3 | <i>F</i> 12.49 (d 10 days); <i>M*</i> 9.51 (d 3 days); M 12.57; M. 1.60 | 1 | 1 | 2 | - | 3 | 1 | 6.23 | 3.16 |
| 4 | <i>F</i> 2.64; <i>F*</i> 3.67 (d 16 days) | - | 1 | - | 1 | - | 2 | 11.43 | 11.38 |
| 5 | <i>M*</i> 3.57 (d 19 days); M 9.58; F 2.60; M 11.62 | 1 | - | 2 | 1 | 2 | 2 | 6.33 | 5.13 |
| 6 | <i>M*</i> 7.67 (d 5 days) | 1 | - | - | - | 1 | - | 9.44 | 4.45 |
| Totals | | 3 | 5 | 5 | 2 | 7 | 8 | | |
| Totals, sexes combined | | 8 | | 7 | | 15 | | | |

Notes: Affected children are in italics. Index patients are marked with an asterisk. Dates following M or F are the month and year of birth. d = died.

All sibs shown are full sibs. In Family 5 there were in addition two half-sibs, one male and one female, both unaffected, both born before the index patient and having the same father as the index patient.

Discussion

In assessing the risk to sibs in this small group of families, if the two index patients in Family 1 were in fact independently ascertained, the proportion of sibs affected is 3 in 10 or 0.3. If the occurrence of the first affected child in Family 1 influenced the referral of the second to the hospital, the family should be counted once only. The proportion of total sibs affected then becomes 2 in 9 or 0.22. Either estimate of the risk to sibs would be compatible with the 0.25 risk to be expected on the assumption of a single autosomal recessive gene. If such recessive inheritance were responsible, then for this rare condition one might expect to find examples of parental consanguinity. In fact in none of the families are the parents known to be related. Also, as far as is known the families are independent of one another, though it is of interest that the same relatively rare surname occurs in two of them, the paternal grandfather (and therefore also the father) in one family having the same surname as the maternal grandfather in another.

Chromosome studies were not made on any of the affected children. Had this condition been associated with a chromosome abnormality, it could be argued that the occurrence of two affected sibs might be related to a chromosomal rearrangement in one or other parent, comparable with the balanced parental translocation t(DqGq) rarely responsible for Down's syndrome in the offspring. This was the reason for making chromosome studies for the parents in Family 1, but in fact the karyotype in each case proved to be normal.

It is clear that the specific type of intestinal malformation known as the 'apple peel' syndrome can recur within a sibship. The present series of families is too small for any accurate estimate of segregation ratios. The recurrence risk to sibs in this series is compatible with the condition being autosomal recessive, but its inheritance may prove to be more complex. Further surveys are needed to establish the exact genetic mechanism involved.

Summary

In a follow-up of an unselected series of children born with the rare form of small bowel atresia known as 'apple peel' deformity, 6 families were studied. In each of 2 families, 2 sibs were affected. The findings are compatible with autosomal recessive inheritance of the condition.

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Addendum

Since this paper was submitted for publication, jejunal atresia has been described in 3 of 16 liveborn sibs in a

single sibship (Mishalany and Najjar, 1968). The family also provides the first recorded instance of parental consanguinity (first cousin relationship) in association with this malformation. The authors suggest that autosomal recessive inheritance is the most likely genetic mechan-

ism, and the present series lends further support to their hypothesis.

REFERENCE

Mishalany, H. G., and Najjar, F. B. (1968). Familial jejunal atresia: three cases in one family. *J. Pediat.*, **73**, 753.