



Familial intussusception

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Abstract

Purpose: Intussusception is one of the common causes of intestinal obstruction in early childhood. Although a genetic predisposition has been suggested in some cases, its etiology is considered to be incidental, and it has not been traditionally regarded as having any genetic basis. Authors report on cases of so-called idiopathic intussusception that demonstrate a strong familial tendency.

Materials: We reviewed medical records of idiopathic intussusception reduced in our institute between 1975 and 2004. There were 564 patients. Their parents and/or grandparents were interviewed directly or by telephone about the occurrence of familial intussusception.

Results: A family history of idiopathic intussusception, including third-degree relatives, was noted on 39 pedigrees. Occurrences between father and son were 1, mother and son 4, mother and daughter 2, mother and son and daughter 1 (sibling), siblings 10, sibling and cousin 1, uncle or aunt and nephew or niece 6, uncle and nephew and niece (cousin) 1, and cousins 13. Among 564 patients with idiopathic intussusception, 20 patients, both of 6 pairs of siblings and both of 4 pairs of cousins were treated in our institute. So in 554 pedigrees of idiopathic intussusception, the incidence of familial intussusception, including third-degree relatives, was estimated to be 7.0% or 1 in 14.2 cases.

Conclusion: We concluded that hereditary predisposition, such as anatomical basis, may be considered as an etiological factor in many cases of idiopathic intussusception. In these predisposed families, if they have inducements such as viral infections as the acquired agents, intussusception will occur very easily. © 2007 Elsevier Inc. All rights reserved.

Intussusception is one of the common causes of intestinal obstruction in early childhood. Although a genetic predisposition has been suggested in some cases, its etiology is considered to be incidental, and it has not been traditionally regarded as having any hereditary basis.

On July 31, 1981, a 3-year-old boy having intermittent abdominal pains was transferred to our institute. A barium enema revealed a so-called idiopathic intussusception. The next day, his 6-month-old younger brother arrived at our institute because he had intermittent abdominal pains. A barium enema showed an idiopathic intussusception. Both

of them were reduced with low hydrostatic pressure. In the siblings, the interval of onset of intussusception was only 28 hours. Sudden appearance of these cases in a short time strongly stimulated our interests in the frequency and etiology of familial intussusception in childhood. The aim of this article is to report our experiences and to discuss the frequency and etiology of this lesion.

1. Materials and methods

We reviewed cases of patients with idiopathic intussusception reduced in our institute between 1975 and 2004. Before July 1981, cases were reviewed retrospectively, and after

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August 1981, a prospective study was performed. During admission and after discharge from our hospital, parents and/or grandparents were interviewed directly or by telephone in detail about the family history of intussusception.

2. Results

2.1. Incidence of familial intussusception

Over a period of 30 years, we treated 564 patients with idiopathic intussusception. A family history of intussusception, including third-degree relatives, was noted in 39 pedigrees. Occurrences between father and son were 1, mother and son 4, mother and daughter 2, mother and son and daughter (sibling) 1, siblings 10, sibling and cousin 1, uncle or aunt and nephew or niece 6, uncle and nephew and niece (cousin) 1, and cousins 13. The family pedigrees of these patients are shown in detail in Fig. 1.

Among 564 patients with idiopathic intussusception, 20 patients (6 pairs of siblings and 4 pairs of cousins in the same family) were also treated in our institute. Therefore, in 554 pedigrees of intussusception, the incidence of familial intussusception including third-degree relatives was estimated to be 7.0% or 1 in 14.2 cases.

2.2. Age at the first episode of familial intussusception

Age at the first episode of familial intussusception in 84 patients varied from 2 months to 4 years 8 months (mean, 14 months). Fifty (59.5%) patients were younger than 12 months, 21 (25%) patients were younger than 24 months, and 13 (15.5%) patients were older than 24 months.

2.3. Interval of the occurrence of intussusception among siblings

The interval of the occurrence of intussusception between siblings was estimated. In case no. 13, it was only 28 hours. In case no. 11, it was 2 months 12 days. In case no. 14, after the first episode of intussusception of a younger sister, her elder brother experienced a fourth episode of intussusception 3 months 26 days later. In the remaining cases, the interval was more than 1 year.

2.4. A case associated with Waugh syndrome (case no. 11)

In July 1977, a boy aged 2 years 4 months was transferred to us because he had vomiting and intermittent abdominal

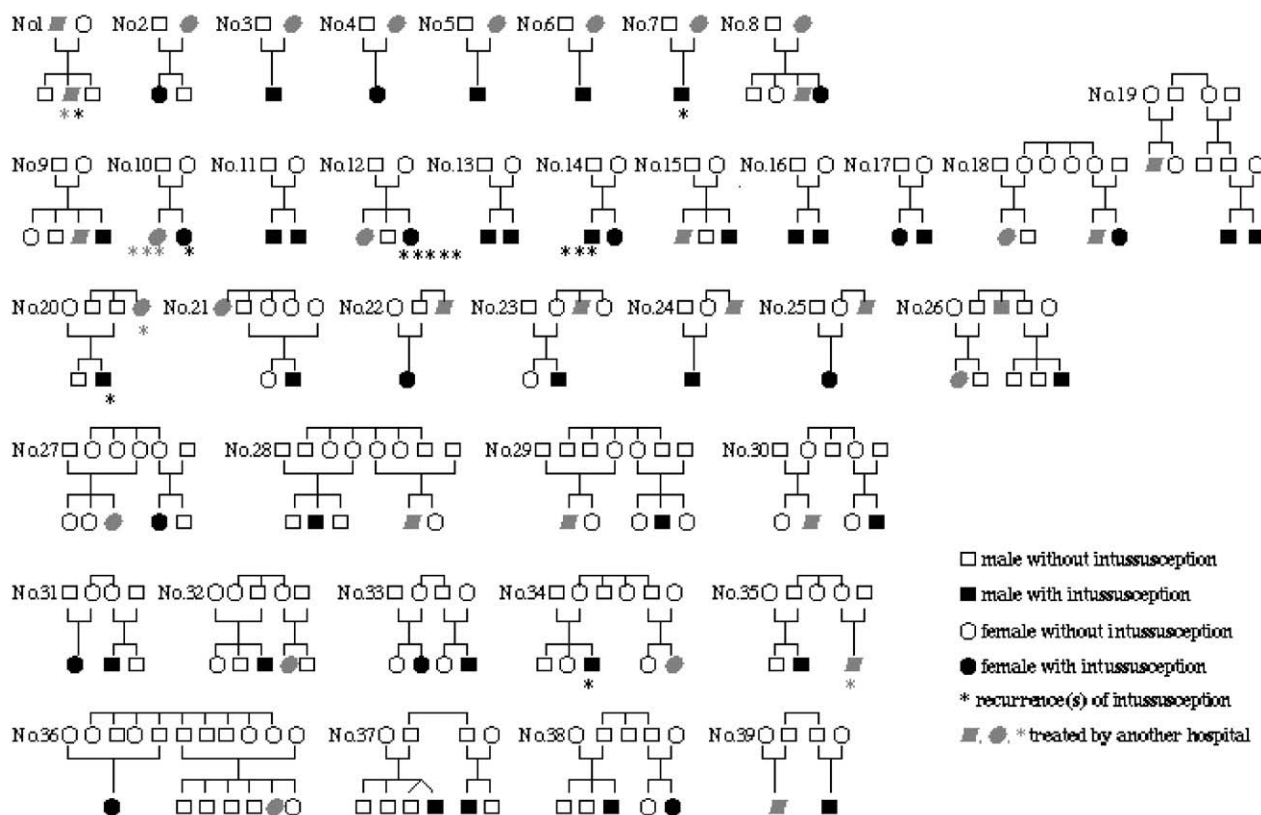


Fig. 1 Pedigrees of familial intussusception.

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