Case report

Coeliac disease in monozygotic twin girls. Synchronous presentation

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SUMMARY A pair of monozygotic twin girls with coeliac disease is reported. The diagnosis was made on clinical and biochemical evidence of malabsorption, characteristic histological findings, and clinical, biochemical, and histological response to gluten elimination. Monozygosity was established on finding a single placenta at birth, exact similarity of physical appearance, similar blood group, and histocompatibility antigens, and negative reaction in mixed lymphocyte culture. This is one of six well documented cases of coeliac disease in monozygotic twins and may throw light on the importance of genetic and environmental factors in the causation and expression of the disease.

The clinical expression and the time of presentation of coeliac disease depend on genetic\(^1\)\(^-\)\(^3\) as well as on environmental\(^4\)\(^-\)\(^5\) factors. Twin studies highlight the importance of both factors, as both discordance\(^6\)\(^-\)\(^8\) and concordance\(^9\)\(^-\)\(^13\) for coeliac disease in monozygotic twins have been reported. The facilities and skills for performing intestinal biopsies has been available to us for the last 2-5 years\(^1\)\(^4\) during which the diagnosis of coeliac disease was made in 14 patients; the area has 10 000–12 000 live births per year. Herein we describe the sixth pair of monozygotic twins concordant for coeliac disease, in whom evidence of monozygosity is shown. The effect of early introduction of gluten is emphasised.

Case report

Two Jordanian girls, HI and SI, were admitted at the age of 18 months because of diarrhoea, irritability, abdominal distension, poor appetite, and failure to gain weight since the age of 6 months. They were born at term to non-consanguinous parents and birth weights were normal (3 kg and 3.5 kg). Both were fed a modified cow’s milk formula from birth. At the age of 2 months a large measure of wheat powder was added to each milk feed.

Examination revealed marked similarity of physical features with gross abdominal distension and wasting. Weights (6.8 kg and 6.7 kg) and lengths (68 cm, 69 cm) were markedly below third percentile for both girls. Bone age was 10 months. Haemoglobin, serum folate, the one hour blood D-xylose, and urinary excretion of D-xylose were all reduced. Sweat chloride, stool examination for parasites, bacteria and reducing substances, and a duodenal aspirate for giardia were negative. Multiple intestinal biopsies obtained during peroral fibreoptic endoscopy\(^14\) showed subtotal villous atrophy compatible with coeliac disease (Figs 1 (1) and 2 (1)).

Histocompatibility antigens were identical: A2, A29, and B8. There was no reaction in the one way or two way mixed lymphocyte culture. Blood group antigens were also similar: A, Rh positive, \(\text{iLe}^a\) negative, \(\text{iLe}^b\) negative, and P positive. No agglutination was detected on direct cross matching between the blood and serum of the twins.

On gluten elimination there was marked acceleration of growth. At the age of 28 months weight and height were on the 25th percentile and weight for height was on the 50th percentile for both girls. By that time there was marked improvement in the biochemical parameters, as well as the histologic appearance of intestinal biopsy (Figs 1 (2) and 2 (2)) in both girls.
Discussion

In the small number of sets of identical twins with coeliac disease so far reported, both concordance\textsuperscript{2-13} and discordance\textsuperscript{6-8} for the disease have been described. It has been estimated that two thirds of monozygotic twins and one third of dizygotic twins are concordant for coeliac disease.\textsuperscript{8} It is then unlikely that a single gene is involved in the pathogenesis of coeliac disease and a multigenes hypothesis is tenable.\textsuperscript{1,8} Furthermore, a specific environmental insult, gluten is essential for the clinical expression of the disease.\textsuperscript{13} Several histocompatibility antigens have been associated with coeliac disease, including B8,\textsuperscript{1} HLA-DW3,\textsuperscript{2} and B-cell antigens.\textsuperscript{3} The two girls in
Coeliac disease in identical twins

Fig. 2  Intestinal biopsies in twin B before (1) and after (2) gluten elimination. (Haematoxylin eosin ×150, original magnification.)

this twin pair had HLA-B8, a genetic marker for coeliac disease. Early and heavy introduction of gluten is the underlying factor in the synchronous clinical expression of the disease. This provides further evidence to the interplay of environmental and genetic factors in the pathogenesis as well as the clinical expression of coeliac disease. Delayed introduction of gluten has been associated with a drop in the incidence of coeliac disease. It may be premature, however, to judge if this is a real drop, or a delay in the clinical expression of the disease in a genetically predisposed population. As in many other conditions, monzygotic twin studies are important in highlighting the influence of genetic and environmental factors in the pathogenesis and expression of coeliac disease.
References


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