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How strict is galactose restriction in adults with galactosaemia? International practice

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ABSTRACT

Dietary management of 418 adult patients with galactosaemia (from 39 centres/12 countries) was compared. All centres advised lactose restriction, 6 restricted galactose from galactosides ± fruits and vegetables and 12 offal. 38% (n = 15) relaxed diet by: 1) allowing traces of lactose in manufactured foods (n = 13) or 2) giving fruits, vegetables and galactosides (n = 2). Only 15% (n = 6) calculated dietary galactose. 32% of patients were lost to dietetic follow-up. In adult galactosaemia, there is limited diet relaxation.

1. Introduction

There has been considerable debate regarding the necessity for severe galactose restriction in older patients with galactosaemia. Patient support groups are posing questions about the essentiality of life-long strict diet and some adults with galactosaemia may already follow a self-relaxed diet. Although poor dietary control beyond infancy has been associated with cataract formation [1,2] and increases in biochemical markers [3], overall life-long dietary restriction does not appear to play a significant role in determining the severity of cognitive outcomes [4–6]. In practice, there is little data about the dietary management of adult patients and many are lost to dietetic follow-up [7]. There are only a few adult case reports of patients homozygous for the severe mutation Q188R who have self-liberated galactose-intake without obvious adverse effects [8–10]. In order to assess the extent of diet relaxation in adult clinical practice, this international survey was conducted to examine dietary advice given to adults with galactosaemia.

2. Materials and methods

2.1. Study design

A questionnaire (17 multiple choice and short answer questions) relating to dietary advice to adult patients with galactosaemia, was sent to dietetic members of the Society for the Study of Inborn Errors of Metabolism (SSIEM) and Australasian Society for Inborn Errors of Metabolism (ASIEM). Dietitians were requested to cascade this questionnaire to other dietetic colleagues within their country. In this cross-sectional audit, data from each clinic was collected about the severity of dietary restriction (quantity of galactose allowed; restriction of galactosides, galactose in fruit and vegetables, offal; and cheese permitted), any dietary changes by patient or dietitian specifically in adulthood, circumstances when dietary treatment might be relaxed, regularity of follow-up; and frequency of galactose-1-phosphate (Gal-1-P) monitoring. Data on patient genotype, long term patient outcome and biochemical data was not included in this audit. Ethical approval was not required as no specific identifiable patient data was obtained.

3. Results

Questionnaires were returned from 39 European and Australasian international IMD centres providing data on 418 adult patients with galactosaemia from 12 countries (Table 1). Each individual centre regularly treated between 1 and 24 adult patients (median: 8 patients) with galactosaemia.

3.1. Overall dietary restrictions

All centres recommended lactose restriction in adult patients, but there was some galactose restriction from fruits and vegetables (4/39) and galactosides (6/39) and 12/39 centres limited offal intake (Table 1). Only one centre in Spain did not permit any low lactose cheese.

3.2. Relaxed diets

In adults, diets were relaxed in 15 centres (38%) from 6 countries (Table 1), but mainly self-directed by patients than dietitians. Centres advising a lactose-free diet only, relaxed diets with trace amounts of lactose in manufactured foods such as breads, biscuits, and cakes (e.g., lactose in sodium and calcium caseinates, animal fats, flavourings) (estimated galactose: <20 mg/portion) or margarine and butter on sandwiches (estimated galactose: up to 200 mg/sandwich) purchased in retail outlets. Centres in France permitted all fruits, vegetables and galactosides and so advised a lactose-free diet only. Although information was not collected about metabolic control, no dietitians reported development of symptoms as a consequence of these small changes.

3.3. Calculation of galactose intake

Most centres (85%, n = 33) did not formally calculate the amount of galactose in the diet. One centre (Belgium) allowed only 50 mg /day, 2 centres (Italy, Netherlands) 100 mg/day, 1 centre (Belgium) 300 mg/day, 1 centre (Germany) 300–500 mg/day, and 1 centre (Germany) 500 mg/day.

3.5. Follow-up

Patients were monitored by dietitians annually by the majority of centres (67%, n = 26). Thirty-two percent (n = 132) of patients were lost to dietetic follow-up (definition: no contact for at least 2 years). Reasons for lack of follow-up included: patient relocation, regular follow-up by physicians but not dietitians, and patient choice only to attend clinics according to need e.g. pregnancy and genetic counselling.

Gal-1-P measurements were monitored by half of the centres (n = 20) annually and 14 (36%) rarely or never monitored this biochemical marker.

4. Discussion

This is the first international multi-centre study to describe dietetic management practices in adult patients with galactosaemia. It is clear that there is uncertainty regarding the requirement for strict diet in adulthood. Although over 75% of centres considered that a strict diet was necessary, lower patient dietary adherence associated with less health professional conviction for the need for strict diet led to a very cautious approach in moderating dietary restriction. Advice in adulthood was also influenced by severity of childhood restriction (lactose free only vs low galactose). To our knowledge no adult suffered from symptoms as a consequence of small dietary changes but there was