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Short report

Consensus on the guidelines for the dietary management of classical galactosemia

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SUMMARY

Background and aim: Worldwide there is scientific discussion about the dietary management of galactosemia. The dietary management is very different in several countries among Europe, the US and Canada. The main points of discussion are related to the fact that i) despite a strict diet some patients still have poor outcomes; ii) there is lack of scientific knowledge about the role of endogenous production of galactose on disease evolution, with or without diet. The aim of the current work was the creation of a Belgian consensus on dietary guidelines for the management of galactosemia.
Methods: A step-wise approach was used to achieve a consensus, including: a workshop, a Delphi round, discussion groups and a round table of different Belgian experts.
Results: The consensus is an agreement between strict guidelines (strict limitation of fruits, vegetables and soybean products/French guidelines) and the more liberal guidelines (comparable with a diet free of lactose/guidelines of UK and the Netherlands). The consensus document consists of different modules, including the medical context, the theoretical background of dietary guidelines and the age-specific practical dietary guidelines.
Conclusion: A Belgian consensus on the guidelines for the dietary management of classical galactosemia was developed despite the uncertainties of the efficacy and practical application of these guidelines. The final consensus is based on scientific knowledge and practical agreement among experts. In the future, regular revision of the guidelines is recommended and a uniform European guideline is desirable.

1. Introduction

Galactosemia is an inherited disorder of galactose metabolism that results from a defect in one of the following enzymes: galactokinase (GALK), GAL-1-phospate uridyltransferase (GALT) and uridine diphosphate-GAL-4-epimerase (GALE). The most common form of galactosemia results from a defect in the GALT activity. Its estimated incidence is 1/40,000–60,000 live births [1]. This form is called the classical galactosemia. Patients with GALT deficiency appear normal at birth but soon develop severe hepatic, renal and gastro-intestinal manifestations which, if not treated, mostly lead to death [2]. Removal of dietary lactose and galactose is essential as this will prevent or decrease the severity of the initial metabolic crisis in the neonate.

Worldwide there is a controversy about the dietary management for galactosemia. A comparative study of Mac Donald et al. shows that the dietary management in all the contributing countries is different [3].

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Meanwhile several European countries developed their own specific dietary management. The United Kingdom published, in 1999, its consensus on dietary guidelines for galactosemia [4]. Other countries published their guidelines later, e.g. the Netherlands in 2005 [5] and France in 2008 [6]. Each national guideline has different opinions on several points (e.g. acceptance of soy-based products, all vegetables allowed or not). This diversity of management plans has been confirmed in a recent publication of Jumbo-Lucioni et al. [7].

The main reasons for variation in dietary guidelines are: i) despite a strict diet, some patients still have poor outcomes. They still develop symptoms of learning difficulties, speaking problems or difficulties in language understanding and movement disorders. Despite strict diet adherence, girls with galactosemia can develop late or no onset of puberty. Patients with galactosemia mostly have fertility problems [8–11]; ii) there is a lack of scientific knowledge about the role of endogenous production of galactose on disease evolution, with or without diet. Endogenous production of galactose is not affected by the exogenous intake from the diet. But galactose tolerance increases with age partly as a result of the age-related decrease of endogenous galactose production. The endogenous production of galactose in adult patients can reach 1000 mg/day while the daily intake of galactose varies between 30 mg and 54 mg, depending on the type of diet [8–10].

Until recently, there was no consensus on the guidelines for dietary management of classical galactosemia in Belgium. The aim of the Belgian metabolic dietitians' workgroup was to develop a Belgian consensus on dietary guidelines for the management of galactosamia. This was an unique process as Belgium has a Dutch speaking and a French speaking community, both having their own specific lifestyle characteristics.

2. Material and methods

Based on the known dietary guidelines worldwide and the different opinions of several countries, the Belgian Inherited Metabolic Disease Centers started to create a national consensus in May 2010. The process took two years. The process consisted of different consecutive steps: a workshop, a Delphi round (requiring up to date knowledge), discussion groups and an expert roundtable were organized. In Fig. 1 a schematic overview of the process is presented.

In the first step, the opinions of the neighboring countries, the Netherlands and France, were presented in parallel with a selected number of other points of view from other countries. The aim of the seminar was to provide all the Belgian experts enough background about common practice in the neighboring countries. This seminar was followed by a roundtable of different Belgian experts (metabolic dietitians and medical doctors, working in the Belgian Inherited Metabolic Disease Centers). A consensus for the different points of discussion was aimed for. The different points of discussion (need for age-specific dietary differences; diet lactose-free or a galactose restricted diet; allowance of soy or not; the use of elemental formula; limitations regarding fruits and vegetables; which cheeses are allowed; allowance of eggs and organ meats; allowance of nuts, seeds and chocolate) were defined upfront, using the results of the seminar, comparing the galactose content of different foods in specialized food composition tables [12] and an extensive literature review available to all participants.

The different discussions and related general decisions were summarized by the two coordinating dietitians in a first Belgian document.

In the next phase a Delphi round was started. The first consensus document was sent out to all ($n = 13$) members of the expert group. New literature and discussions between metabolic experts (doctors and dietitians) led to further adaptations of the document. These adaptations were included in a second version of the document. The second version of the consensus document was discussed in a new round table of all Belgian metabolic dietitians, resulting in a third document. This version was finally sent out to all members of the expert group. All remarks were summarized by the two coordinating dietitians in a final document.

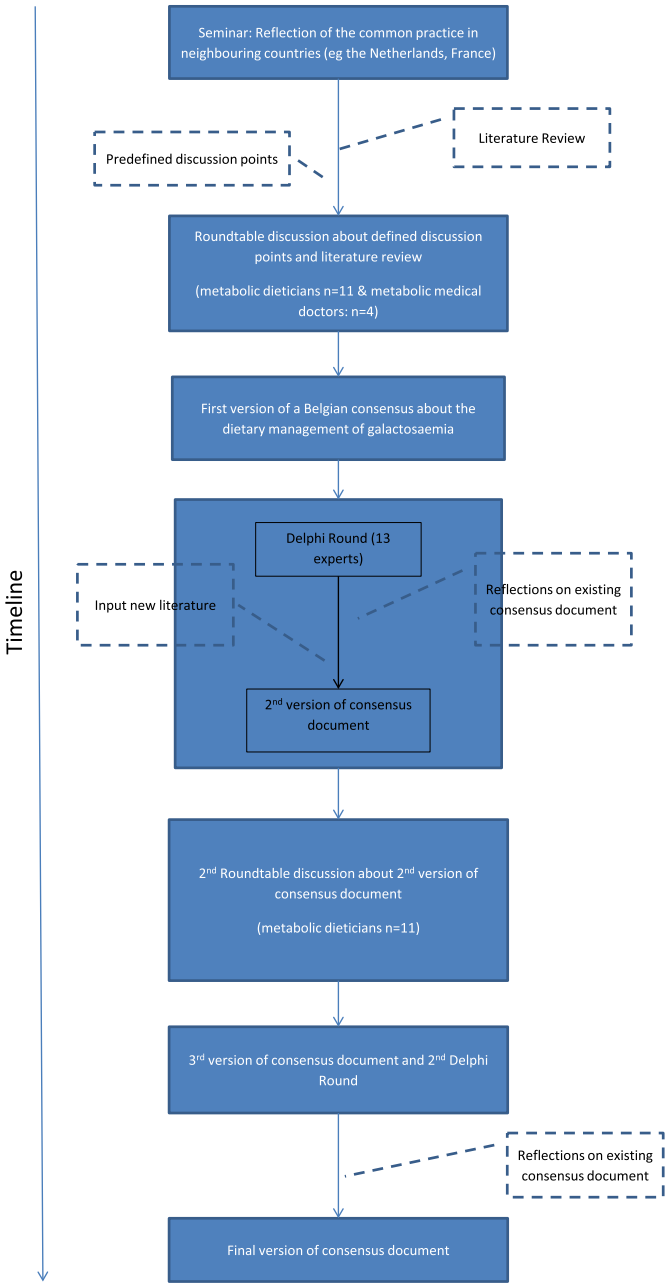

Fig. 1. Decision making process and graphical presentation of used methodology.

On the annual meeting of Belgian metabolic experts "Metabolics.be", May 2011, "The Dutch version of the Belgian consensus" was presented. In a last step the consensus was translated into French.

3. Results & discussion

Knowing the current ambiguity of the dietary management of galactosamia regarding the optimal strictness and duration of dietary galactose restriction and the need for evidence-based best