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Dietary practices in propionic acidemia: A European survey

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ABSTRACT

Background: The definitive dietary management of propionic acidaemia (PA) is unknown although natural protein restriction with adequate energy provision is of key importance.
Aim: To describe European dietary practices in the management of patients with PA prior to the publication of the European PA guidelines.
Methods: This was a cross-sectional survey consisting of 27 questions about the dietary practices in PA patients circulated to European IMD dietitians and health professionals in 2014.
Results: Information on protein restricted diets of 186 PA patients from 47 centres, representing 14 European countries was collected. Total protein intake [PA precursor-free L-amino acid supplements (PFAA) and natural protein] met WHO/FAO/UNU (2007) safe protein requirements for age in 36 centres (77%). PFAA were used to supplement natural protein intake in 81% (n = 38) of centres, providing a median of 44% (14–83%) of total protein requirement. Seventy-four per cent of patients were prescribed natural protein intakes below WHO/FAO/UNU (2007) safe levels in one or more of the following age groups: 0–6 m, 7–12 m, 1–10 y, 11–16 y and > 16 y. Sixty-three per cent (n = 117) of patients were tube fed (74% gastrostomy), but only 22% received nocturnal feeds.
Conclusions: There was high use of PFAA with intakes of natural protein commonly below WHO/FAO/UNU (2007) safe levels. Optimal dietary management can only be determined by longitudinal, multi-centre, prospective case controlled studies. The metabolic instability of PA and small patient cohorts in each centre ensure that this is a challenging undertaking.

1. Introduction

Propionic acidaemia (PA, OMIM #606054) is a rare, life threatening, inherited metabolic disorder with a poor clinical outcome [1]. Movement disorders, cardiomyopathy, prolonged QT, thrombocytopenia and pantocytopenia are common clinical manifestations [2]. PA is caused by deficiency of propionyl-CoA carboxylase (PCC; E.C. 6413) which catalyses the carboxylation of propionyl-CoA to D- methylmalonyl-CoA [3]. Propionyl-CoA is derived from three main sources: 1) catabolism of isoleucine, methionine, valine and threonine, 2) odd-chain fatty acid metabolism and 3) bacterial fermentation of carbohydrate in the gut [4].The main principles of management in PA are to minimise the production of toxic metabolites of organic compounds whilst supporting anabolism, normal growth and good nutritional status. Dietary treatment, together with carnitine and antibiotics are important components of management [5].

In 2014, proposed European guidelines were published on the diagnosis, acute and chronic management of PA [5]. They were developed using SIGN methodology, based on a critical appraisal of all scientific evidence. There were three main dietary recommendations: 1) there should be an adequate energy supply combined with avoidance of prolonged fasting, 2) lower intake of precursor containing amino acids by restricting natural protein intake, and 3) use of PA precursor-free amino acid supplements (PFAA) only if natural protein tolerance is below the WHO/FAO/UNU (2007) safe levels of protein intake [6].

The PA clinical guidelines aimed to improve the consistency of care and provide authoritative recommendations and consensus to reassure practitioners about the appropriateness of treatment practices. In other inherited metabolic conditions, it has already been established that dietary treatment varies widely and is influenced more by geographical region of care [7,8] than disorder severity [9]. The PA dietary guidelines are based on low grade scientific evidence (mainly level D) and it is possible that these treatment guidelines may have little impact on individual practice.

In order to examine dietetic practices in PA, in a cross-sectional survey we have collected data from health professionals across Europe immediately prior to the publication of the PA guidelines [5].

2. Material and methods

In July to August 2014, a questionnaire (including 27 multiple choice or short answer questions) about 3 types of organic acidaemia's (isovaleric acidaemia [IVA], propionic acidaemia [PA] and methylmalonic acidaemia [MMA]) was sent to all European members of the Society for the Study of Inborn Errors of Metabolism Dietitians Group (SSIEM-DG) and other European dietitians (n = 53) who have previously participated in surveys [7,9]. They were requested to cascade this questionnaire to dietitians and physicians within their own country. The IVA results have been previously published [8]. All the questions were written in English. Information on patient numbers,

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