Clinical pathways for inborn errors of metabolism: warranted and feasible

Serwet Demirdas1†, Imke N van Kessel1†, Marjolein J Korndewal1, Carla EM Hollak2, Hanka Meutgeert3, Anja Klaren3, Margreet van Rijn4, Francjan J van Spriens4, Annet M Bosch1* and Dutch working Group

Abstract
Inborn errors of metabolism (IEMs) are known for their low prevalence and multidisciplinary care mostly founded on expert opinion. Clinical pathways are multidisciplinary tools to organise care which provide a clear route to the best care and improve communication. In 2010 the Dutch Society for Children and Adults with an Inborn Error of Metabolism (VKS) initiated development of clinical pathways for inborn errors of metabolism. In this letter to the editor we describe why it is warranted to develop clinical pathways for IEMs and shortly discuss the process of development for these pathways in the Netherlands.

Keywords: Phenylketonuria, PKU, Clinical pathway

Introduction
Inborn errors of metabolism (IEMs) are known for their low prevalence and chronic need of medical care. Care provided is multidisciplinary and often based on expert opinion. In recent years, excellent guidelines on metabolic disorders have been developed and published [1-5]. In 2010 the Dutch Society for Children and Adults with an Inborn Error of Metabolism (VKS) initiated development of clinical pathways for 20 IEMs, with separate versions for professionals and patients. This letter discusses why clinical pathways for IEMs are warranted and feasible.

Background
Clinical pathways are a tool for multidisciplinary decision making and organization of care processes for well defined groups of patients [6]. Often they are based on guidelines [7-9]. Pathways optimize clinical outcomes whilst maximizing clinical efficiency [10]. For example, they describe which actions should be taken, when, and by whom [11]. It has been demonstrated that use of pathways decreases duration of inpatient care, increases interdisciplinary communication, enhances patient knowledge and self awareness, leads to significant better coordination of care and reduces costs [7,9,12-14].

Clinical pathways can be valuable for patients with IEMs. Firstly, low prevalence of IEMs leads to limited knowledge about best practice. In the absence of robust evidence, expert opinion and outcomes of clinical studies can support the establishment of a clinical pathway [12]. When frequently updated, it presents a reference to latest state of art in care [15,16] and provides guidance for further research. Secondly, a multidisciplinary approach is of great importance. The complexity of multidisciplinary care may lead to delay of care, overuse of diagnostics or therapy and miscommunication between caregivers [7]. Multidisciplinary cooperation using a clinical pathway will improve communication and provide a clear route to best care, based on consensus. Thirdly, clinical pathways may improve care for patients when used in local hospitals, while the physicians in academic referral centers can serve as consultants. Finally, clinical pathways become more important as transition to adult care increases [17], leading to more active participation of patients in their treatment.

The design of clinical pathways for inborn errors of metabolism

Design and consensus
The initiative for development of clinical pathways was taken by the patient society (VKS). Dutch expert
pediatricians, internists and dieticians for each specific disorder in cooperation with the VKS created the pathways. The final version was discussed in a national consensus meeting. Separate versions were made for professionals and for patients, presenting the Dutch consensus. All advice is substantiated by a level of evidence [18], according to the scoring system of the Dutch Institute for Healthcare Improvement CBO. Level 1: 1 systematic review or 2 independent high quality randomized controlled trials (RCTs); level 2: 2 independent moderate RCTs or comparative trials; Level 3: 1 RCT, comparative or non-comparative trial; Level 4: expert opinion [19].

Clinical pathway for professionals
The first section of the version for professionals comprises a general introduction and concise strategy for diagnostics and treatment. Second and third sections contain more specific guidance for treatment and follow up in childhood and adulthood.

The pathways include responsibilities for each professional, advised frequency for outpatient visits and laboratory studies, and recommendations on follow up of known complications of the disorder. In the pediatric pathway one chapter is dedicated to transition from pediatric to adult care.

In the pathways all advice is substantiated by a level of evidence. Evidence levels 3 and 4 were common. Level 1 was rarely available and mostly resulted from trials evaluating a novel pharmaceutical agent. Most advice was therefore founded on expert opinion and trials of moderate quality.

Clinical pathway for patients
The first section of the patient version contains general information on the disorder and its treatment. The second and third sections address treatment and follow up in childhood and adulthood. The purpose of the pathway for patients is to provide insight into current consensus of best practice and an overview of all professionals involved. It provides clarity on responsibilities, including that of the patient/parents who take a prominent place in the treatment team.

For active patient participation, patients must be provided evidence based information in an appropriate and comprehensible form [20]. The fact that the patient versions are based on the professional pathway ensures that they are in accordance with available evidence, and comprehensibility is secured by cooperation with the VKS.

We demonstrated that development of clinical pathways for IEMs is feasible and we were able to reach national consensus. At this time, Dutch pathways are publically available for 20 diseases including urea cycle defects, organic acidurias, mitochondrial fatty acid oxidation disorders, galactosemia, phenylketonuria, tyrosinemia, glycogen storage disorders, congenital disorder of glycosylation type 1a, and Niemann Pick type c [21].

Abbreviations
VKS Dutch society for children and adults with an inborn error of metabolism; IEMs: Inborn errors of metabolism; PKU: Phenylketonuria.

Competing interests
None of the authors, or any of the members in the working group, have financial or non-financial competing interests to declare.

Authors’ contributions
SD has made substantial contributions to conception and design, has been involved in drafting of the manuscript and has given final approval of the version to be published. INH has made substantial contributions to conception and design, has been involved in drafting of the manuscript and has given final approval of the version to be published. MIK has made substantial contributions to conception and design, has been involved in drafting of the manuscript and has given final approval of the version to be published. CEMH has been involved in drafting of the manuscript and has given final approval of the version to be published. ARB has made substantial contributions to conception and design, has been involved in drafting of the manuscript and has given final approval of the version to be published. AK has been involved in drafting of the manuscript and has given final approval of the version to be published. MR has been involved in drafting of the manuscript and has given final approval of the version to be published. FJS has been involved in drafting of the manuscript and has given final approval of the version to be published. AMB has made substantial contributions to conception and design, has been involved in drafting of the manuscript and has given final approval of the version to be published. INK has made substantial contributions to conception and design, has been involved in drafting of the manuscript and has given final approval of the version to be published. All members of the The Dutch working Group on clinical pathways for inborn errors of metabolism have given final approval of the version to be published.

Authors’ information
Dutch working Group on clinical pathways for inborn errors of metabolism. Folkert W. Asselbergs1, Christiaan Blank2, Terry G.J. Derks3, Eugene F. Dietman4, Monique E. Dijsselhof5, Marc Engelen6, Peter M. van Hasselt7, Nienke M. ter Horst8, Dorine A.M. van den Hurk9, Mirjan C.H. Janssen10, Francois P.J. Kastens11, Elles van der Louw12, Eva Morava13, Joost Nicol14, Ludo van de Pol15, Bwue Tien Poll-The16, Estella Rubio-Gozalbo17, G. Peter A. Smit18, Jessica de Ruijter19, Corrie Timmer20, Catharina M.L. Towe21, Gepke Visser22, Harold W. de Vaal23, Frits A. Wijburg24, Monique Williams25, Departments of Cardiology26, Pediatrics27, Dietetics28 and Internal Medicine29, University Medical Center, Utrecht, The Netherlands. Department of Pediatrics30, Neurology31, Emma Children’s Hospital, Academic Medical Center, Amsterdam, The Netherlands. Section of metabolic diseases32, Beatrix Children’s Hospital, University Medical Center Groningen, University of Groningen, Groningen, The Netherlands. Department of Internal Medicine33 and Pediatrics34, University Medical Center St Radboud, Nijmegen. Department of Internal Medicine35, Dietetics36, Neurology37, Emma Children’s Hospital, Academic Medical Center, Amsterdam, The Netherlands. Section of metabolic diseases38, Beatrix Children’s Hospital, University Medical Center Groningen, University of Groningen, Groningen, The Netherlands. Department of Internal Medicine39 and Pediatrics40, Erasmus MC, Rotterdam, The Netherlands. Department of Neurology41 and Pediatrics42, University Hospital Maastricht, Maastricht, The Netherlands.

Author details
1Department of Pediatrics, Emma Children’s Hospital, Academic Medical Center, University of Amsterdam, Amsterdam, The Netherlands. 2Department of Internal Medicine, Division of Endocrinology and Metabolism, Academic Medical Center, University of Amsterdam, Amsterdam, The Netherlands. 3Department of Internal Medicine, Division of Endocrinology and Metabolism, Academic Medical Center, University of Amsterdam, Amsterdam, The Netherlands. 4Department of Internal Medicine, Division of Endocrinology and Metabolism, Academic Medical Center, University of Amsterdam, Amsterdam, The Netherlands. 5The Dutch Society for Adults and Children with an Inborn Error of Metabolism (VKS), Zwolle, The Netherlands. 6Division of Metabolic Diseases, Beatrix Children’s Hospital, University Medical Center Groningen, University of Groningen, Groningen, The Netherlands.

Received: 10 December 2012 Accepted: 21 February 2013
Published: 25 February 2013
Volwassenen, Kinderen en Stofwisselingsziekten; Zorgpaden voor

References


20. Coulter A: Evidence based patient information. is important, so there needs to be a national strategy to ensure it. BMJ 1998, 317:225–226.


Submit your next manuscript to BioMed Central and take full advantage of:

• Convenient online submission
• Thorough peer review
• No space constraints or color figure charges
• Immediate publication on acceptance
• Inclusion in PubMed, CAS, Scopus and Google Scholar
• Research which is freely available for redistribution

Submit your manuscript at www.biomedcentral.com/submit