Clinics and Research in Hepatology and Gastroenterology (2015) 39, 205—214

Available online at ScienceDirect www.sciencedirect.com

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ORIGINAL ARTICLE

The quality of hereditary haemochromatosis guidelines: A comparative analysis

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Available online 23 October 2014

Summary

Background and objectives: Hereditary haemochromatosis (HH) is the most prevalent genetic liver disease, with an incidence of 1/200 to 1/400 in the Caucasian population. HH patients are treated by family physicians as well as different specialists. When left untreated or insufficiently

Abbreviations: COPD, Chronic obstructive pulmonary disease; HH, Hereditary haemochromatosis; AGREE II, Appraisal of Guidelines, Research and Evaluation II; EASL, European Association for the Study of the Liver; AASLD, American Association for the Study of Liver Diseases; DUTCH, Netherlands Association of Internal Medicine (NIV), the Netherlands Society of Clinical Chemistry and Laboratory Medicine (NVKC) and Association of Laboratory Physicians (VAL); GRADE, Grades of Recommendation, Assessment, Development, and Evaluation system; CBO, Dutch Institute for Health Care Quality; ICC, Intraclass correlation; AST, Aspartate aminotransferase; ALT, Alanine aminotransferase; TS, Transferrin saturation; SF, Serum ferritin; LI, Liver iron; DM, Diabetes mellitus; PCT, Porfyria cutanea tarda; α-Fp, Alfa-fetoprotein; ECG, Electrocardiogram; TSH, Thyroid-stimulating hormone; DEXA, Dual Energy X-Ray Absorption; MRI, Magnetic resonance imaging; R, Recommended; NR, Not recommended; PR, Partially recommended; NM, Not mentioned.

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http://dx.doi.org/10.1016/j.clinre.2014.09.001

treated, the complications can become life threatening. To support and evaluate qualitative care for HH, we evaluated and compared the available structured guidelines on screening, diagnosis and management of HH patients.
Methods: Seven appraisers systematically reviewed the retrieved guidelines. The Appraisal of Guidelines Research and Evaluation II (AGREE II) was used to score and discuss the quality and reach consensus. The content of recommendations and the evidence behind them, were evaluated.
Results: Three guidelines, developed by the American Association for the Study of Liver Diseases (AASLD), the European Association for the Study of the Liver (EASL) and a DUTCH guideline were found. Fifty-seven percent of the recommendations were not shared between the guidelines, pointing to inconsistency of their content. Only two references supporting the recommendations were shared between all three guidelines. The AASLD guideline contains no information about management and follow-up. Moreover, the methodological quality of the AASLD guideline was rated insufficient, except for 'clarity and presentation' (77%). Applicability of the guidelines was scored very low in all three (AASLD: 31%, EASL: 23%, DUTCH: 35%). The DUTCH guideline was judged best.
Conclusions: Very poor consistency between available guidelines for HH hampers qualitative care and its evaluation. An updated high-quality and evidence-based guideline that covers follow-up and management of patients with HH is needed.

Introduction

Clinical practice guidelines are developed to improve efficacy, to reduce inappropriate practice [1] and to bridge the gap between research and practice [2]. The unrestricted availability of online databases as Medline and Embase should help to establish international consensus on evidence to support recommendations for clinical care, emanating in guidelines [3,4].

Several studies in chronic care, for example in the field of chronic obstructive pulmonary disease (COPD), show that guidelines can improve patient outcomes as well as the care process [5,6]. However, there is an increasing concern about variations of guideline quality and recommendations [7—9].

Guidelines on the same topic, especially when evidence is weak, often differ. Reasons for differences in recommendations are lack of sufficient evidence [10,11], difference in interpretation of evidence [12,13], unsystematic guideline development methods [14,15] and cultural factors [16,17].

Hereditary haemochromatosis (HH) is an autosomal recessive disorder, with a genetic prevalence of 1/200 to 1/400. It is very common and has an estimated carrier frequency of 1/10 in those from Northern European descent. The phenotype results from inappropriate accumulation of iron, resulting in end-organ damage [18]. Symptoms can be absent, but may also be debilitating; complications of the disease can be life threatening, as are diabetes mellitus, osteoporosis, cirrhosis, hepatocellular carcinoma [19]. The varying criteria for case definition, referral, diagnosis, interpretation of test results, follow-up, family screening approaches may lead to confusion in diagnosis and orientations for physicians, patients and their relatives.

To support and evaluate qualitative care for HH, we studied the consistency in recommendations in the available guidelines, with emphasis on recommendations for treatment, follow-up, detection and management of complications of iron accumulation and the scientific evidence supporting these recommendations.

We examined guidelines with a focus on three aspects: (1) the methodological quality of each guideline, examined with the international AGREE II (Appraisal of Guidelines for Research and Evaluation) instrument [20]; (2) the content of guideline recommendations; and (3) the use of evidence.

Methods

Selection of guidelines

We searched for references to guidelines on HH in the Medline database as well as the National Guideline Clearinghouse using following MESH terms: 'haemochromatosis' and 'practice guideline (publication type)' or 'practice guidelines as topic' (June 2013). We included only evidence-based guidelines with clearly defined recommendations from the last 10 years. If the guideline had been updated, the latest version was used.

We included three guidelines from different professional organizations: European Association for the Study of the Liver (EASL), American Association for the Study of Liver Diseases (AASLD) and Netherlands Association of Internal Medicine (NIV), the Netherlands Society of Clinical Chemistry and Laboratory Medicine (NVKC) and Association of Laboratory Physicians (VAL) (DUTCH) [21—24].

Participants

We established an expert panel of seven experts from five disciplines (two hepatologists, two internists, one general practitioner, one laboratory physician and one researcher)