Health Policy 105 (2012) 110–118

Contents lists available at SciVerse ScienceDirect

Health Policy

journal homepage: www.elsevier.com/locate/healthpol

A framework to evaluate the effects of small area variations in healthcare infrastructure on diagnostics and patient outcomes of rare diseases based on administrative data

Tom Stargardt*, Jonas Schreyögg

Hamburg Center for Health Economics, University of Hamburg, Esplanade 36, 20354 Hamburg, Germany

ARTICLE INFO

Article history:
Received 18 October 2011
Received in revised form 19 January 2012
Accepted 23 January 2012

Keywords:
Rare diseases
Administrative data
Healthcare system
Small area variations

ABSTRACT

Introduction: Small area variations in healthcare infrastructure may result in differences in early detection and outcomes for patients with rare diseases.
Methods: It is our aim to provide a framework for evaluating small area variations in healthcare infrastructure on the diagnostics and health outcomes of rare diseases. We focus on administrative data as it allows (a) for relatively large sample sizes even though the prevalence of rare diseases is very low, and (b) makes it possible to link information on healthcare infrastructure to morbidity, mortality, and utilization.
Results: For identifying patients with a rare disease in a database, a combination of different classification systems has to be used due to usually multiple diseases sharing one ICD code. Outcomes should be chosen that are (a) appropriate for the disease, (b) identifiable and reliably coded in the administrative database, and (c) observable during the limited time period of the follow-up. Risk adjustment using summary scores of disease-specific or comprehensive risk adjustment instruments might be preferable over empirical weights because of the lower number of variables needed.
Conclusion: The proposed framework will help to identify differences in time to diagnosis and treatment outcomes across areas in the context of rare diseases.

1. Introduction

The provision of adequate care for rare diseases, i.e. diseases with prevalence below 5/10,000 inhabitants [1], poses additional problems compared to common diseases. The low prevalence and the high number of different rare diseases create a complex and demanding environment within which physicians must detect possible symptoms and find suitable treatment alternatives. Because of their low prevalence, the disease-specific combinations of symptoms are not part of the physician's daily diagnostic and therapeutic activities. This is why rare diseases are often not detected under routine care conditions: diagnosis and treatment of rare diseases thus requires a higher degree of knowledge and specialization.

It is often assumed that the existence of an excellent healthcare infrastructure in a given area is likely to improve diagnostic quality, thereby allowing early detection of the disease and so improving long-term prognosis. With respect to various common diseases, there is a long tradition – stretching back to the 1970s – of research on small area variations, i.e. differences in the rates of use of medical services, especially regarding practice patterns [2,3]. Phelps found that small area variations are likely to be associated with welfare losses [4]. However, the effects of small area variations in healthcare infrastructure on early detection and health outcomes for rare diseases have so far not been evaluated.

A review of existing literature revealed that several methodological aspects have been dealt with separately

* Corresponding author. Tel.: +49 40 42838 8044; fax: +49 40 42838 8043.
E-mail address: tom.stargardt@wiso.uni-hamburg.de (T. Stargardt).

0168-8510
doi:10.1016/j.healthpol.2012.01.011

and only in the context of common diseases. It is thus the aim of this article to provide a comprehensive framework for evaluating small area variations in healthcare infrastructure on the diagnostics and health outcomes of rare diseases. As rare diseases are often complex, chronic and require integrated care across sectors, studies must be able to follow patients along the care pathway over time. Therefore, we focus on administrative data. It allows for relatively large sample sizes even though the prevalence of rare diseases is very low. It also makes it possible to link information on the availability of healthcare infrastructure to data on morbidity, mortality, and healthcare utilization across different sectors.

Following the introduction, in Section 2 we outline study designs that allow us to measure the time between the first symptom of a rare disease and its first diagnosis. We structure this section along the main challenges for defining the study design. We describe different methods for identifying patients with a rare disease or its symptoms in administrative data through the use of diagnoses, procedure codes, reimbursement codes, ATC codes, or a combination of those, and taking into account data quality. We also describe the study setting to analyse delay in diagnosis and treatment outcomes and present a conceptualization of healthcare infrastructure within the ambit of this research. Section 3 addresses the adaption of disease-specific or comprehensive risk adjustment methods to rare diseases using administrative data, e.g. the Charlson Comorbidity Index or the Elixhauser Score. Section 4 proposes statistical methods to measure the effect of healthcare infrastructure on diagnostics and outcomes in the context of rare diseases. The final section addresses possible health policy implications based on studies which use this framework.

2. Choice of study design

Because of their low prevalence as well as their high complexity, rare diseases present a challenge not only for the healthcare system, but also for research based on administrative data. From the researcher's viewpoint, the rarity may result in a low number of observations. Thus, any administrative database accessed for retrospective analysis needs to be large. We propose the following study design based on administrative data for evaluating whether special regional characteristics facilitate early diagnosis of rare diseases or improve outcomes after treatment.

2.1. Identification of a patient with a rare disease in administrative data

The identification of patients in an administrative database is usually accomplished by searching for diagnoses related to in- or outpatient care episodes that identify the disease according to the International Classification of Diseases [ICD]. Ideally, this would require a prior validation of the criteria with respect to the database that is used [5]. This is because the reliability of coded ICDs primarily depends on the coding requirements imposed on the providers by the healthcare system, as well as the variability in disease coding systems themselves, i.e. ICD-10 vs. ICD-9, and on the data-handling process between physician and database [6].

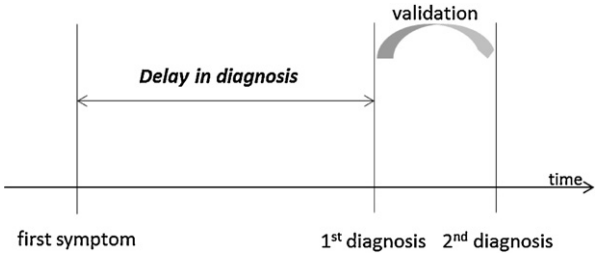

Fig. 1. Measuring delay in diagnosis.

If detailed coding is a prerequisite for reimbursement, as it is in case of inpatient hospital stays reimbursed through DRGs [7], the occurrence of a single ICD code for a disease can be considered sufficient to identify a patient [8]. If detailed coding is not a prerequisite for reimbursement, as for example in the case of outpatient physician contacts reimbursed through lump sum payments, at least one further diagnosis relating to the disease within 180 days or 360 days should be required to validate the first diagnosis. Rare diseases are often hidden in ICD codes of other rare or common diseases. In such cases, researchers have to look carefully for alternatives and may have to combine different ICD codes. Combinations of ICD codes and prescription records or – if available – procedure codes can also be used and might outperform identification by ICD only [6,9]. In any case, for very rare diseases, a combination of different classification systems has to be used due to multiple diseases sharing one ICD code. For some diseases, e.g. osteonecrosis [10], the use of administrative databases alone is not sufficient to identify patients because the respective ICD codes covers many similar diseases and researchers can only identify the disease when additional information from clinical records is taken into account. Thus administrative databases may have to be combined with a review of medical records. It should also be considered that for some rare diseases undercoding and resulting reporting bias may be a problem, because physicians may not know or use the appropriate codes for certain rare diseases.

2.2. Study setting to analyse delay in diagnosis and treatment outcomes

Delay in diagnosis can be further differentiated into delay by patients, i.e. the time between the onset of symptoms and time of first medical contact, and delay by providers, i.e. the time between the first medical consultation and start of treatment [11]. Delay by patients cannot be observed if administrative data alone is used. Once a patient with a rare disease has been identified, however, delay by providers can be best approximated by the number of days between the occurrence of the first symptom that is characteristic for the disease in the database and the date the patient is diagnosed (see Fig. 1). For most rare diseases, the date of the first diagnosis will also mark the start of treatment, or treatment will at least start shortly afterwards.