



Prevalence of rare diseases in the ambulatory health care sector in Germany 2008-2011

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Abstract

Background:

Rare diseases are a very heterogeneous group of medical conditions. The only thing they definitely have in common is their rarity. Rare diseases are mostly inherited and life-threatening or chronically debilitating diseases that affect by definition less than 50 people per 100,000 inhabitants. Up to now about 8,000 different rare diseases are known and due to sophisticated biomolecular knowledge it is very likely, that this number will keep increasing. In Germany more than 4 million people are affected by one of those known rare diseases.

Though there is a growing public awareness concerning rare diseases, these special medical conditions are still insufficiently explored and therefore underdiagnosed and often undertreated. This aspect is the starting point of the following study.

Method:

For estimating the prevalence of different rare diseases in Germany, the ambulatory health care claims data from 2008 to 2011 were used.

At the beginning, the most challenging aspect of the analysis was the reduction of eligible rare diseases to a manageable quantity. Therefore, in the first place we used the Orphanet report on the prevalence of rare diseases from May 2014. All conditions that were neither infectious disease nor related to oncological conditions with unambiguous ICD-10GM Code and a prevalence rate higher than 3 per 100,000 were included. Based on that procedure we were able to identify 88 different rare diseases.

Patients were included, if they were assuredly diagnosed with the same disease in two different quarters of one year. For estimating the prevalence of every single condition, we used the average number of patients per year during the investigation period. This approach reduced the influence of annual variation in the number of cases and allowed a stable estimation of prevalence. Otherwise, due to the small number of affected people per condition, already small variations in the number of cases might have had a huge influence on annual prevalence rates. The reference population (denominator) for prevalence estimation was defined by the total number of statutory health care insured persons.

Results:

Within the study period an average of 573,520 diagnosed patients per year were identified. This equates to an overall prevalence of almost 1% (about 824 diagnosed patients per 100,000 statutory health care insured inhabitants). Please keep in mind that the reported overall prevalence just refers to the analyzed rare diseases and is much lower than the prevalence of all known rare diseases. Having a look

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at single entities, a huge variation in prevalence becomes obvious. The estimated prevalence rates varied from 0.01 patients per 100,000 inhabitants (n=8) describing the Craniorachischisis, the most severe form of neural tube defect, to 112.6 patients per 100,000 inhabitants (n=78,372) on Menière disease, which is a chronic idiopathic inner ear disorder. Additionally, regional variation in prevalence rates was analyzed for some diseases. The results were quite interesting: for example the prevalence for acute sarcoidosis ranged from 32/100,000 in Hesse to 69/100,000 in Mecklenburg-West Pomerania.

The quality of results was evaluated recording to the results of the nationwide neonatal screening and showed a sufficient congruence.

Conclusion:

This study is the first widespread analysis of the prevalence of rare diseases using ambulatory health care claims data and therefore contributes to a growing knowledge and better understanding of rare diseases. Despite methodical challenges, reliable prevalence data was gathered. Perspectively, this study could be or even should be the basis for further research with a focus on patient centered care. Thereby currently unused potentials for improvement of care could be identified and assigned to regular ambulatory care.

Keywords:

Rare diseases, prevalence, sarcoidosis , neonatal screening, ambulatory health care claims data, regional variation

Citation:

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