REAL-WORLD HEALTHCARE RESOURCE UTILIZATION AND RELATED COSTS ASSOCIATED WITH RARE DISEASE IN THE BRAZILIAN PRIVATE MARKET

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BACKGROUND

Patients with rare diseases (RD) face significant challenges from diagnosis through the treatment phase. Furthermore, the extent of the impact of this group of diseases in the Brazilian private market is not well understood.

Due to the rarity and fragmentation of patients across thousands of different diseases, the medical needs of RD patients are not well-defined or quantified within healthcare systems. However, estimates indicate that, collectively, RDs affect 6 to 8% of the population, which suggests they should be recognized as a priority in healthcare policies.

As a result of prolonged diagnostic journeys, RD patients tend to be significant users of healthcare resources. Some estimates suggest that the related costs associated with RD patients may account for over 10% of total healthcare costs.

OBJECTIVES

Rare diseases patients face health challenges and limited treatments, and their impact within Brazilian healthcare is unknown. This study investigated healthcare resource utilization (HCRU) to provide a better understanding of economic impacts of RD.

METHODS

We developed a new methodology starting from a set of rare disease diagnostic codes established by the Orphanet consortium. Based on this list, we matched the equivalent ICD-10 codes within our information system. In a second phase, we excluded conditions with a prevalence of 65 per 100,000 or higher in our population, resulting in 793 ICD-10 codes used in this study. For comparative purposes, we defined another group of individuals with common conditions (CCs), which included any valid ICD-10 code not listed in this final RD list.

From January/2019 to December/2023

793 ICD-10 codes linked as RDs

This retrospective cohort study included all beneficiaries who had a record in the health plan's administrative database with one of the 793 ICD-10 codes listed in any diagnostic field and a healthcare utilization date between January 1, 2019, and December 31, 2023.

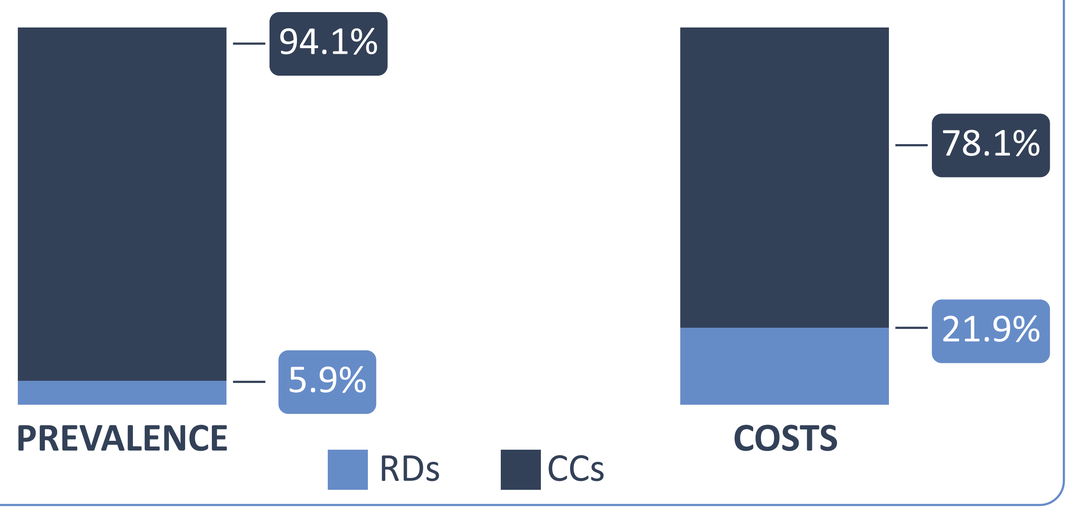
Direct medical costs were calculated annually for patients with costs independent of the stage of diagnosis. Data were aggregated by prevalence, costs, and demography. Chi-square and Fisher's exact and Student's T-tests for categorical/continuous measures were used. Statistical significance for $p<0.05$.

RESULTS

A total of 60,012 beneficiaries were analyzed, with an average age of 53 years. When combined (Figure 1), rare diseases collectively affected 5.9% of the population (n=3,564; mean age 63.4 years; 61.4% female). The prevalence of rare diseases was significantly higher in women (6.1%) than in men (5.6%) ($p=0.013$).

Regarding healthcare expenses over the 5-year study period, these totaled slightly over $367 million in health plan costs (Figure 1), with 21.9% ($80 million) attributed to rare diseases and $287 million to common conditions, despite rare disease patients representing only a small percentage of the total population.

Figure 1 – Rare diseases and common conditions: prevalence and respective costs



The average healthcare expenditure per patient (Figure 2) was nearly four times higher for rare diseases ($6,816) compared to common conditions ($1,960) ($p<0.001$).

Figure 2 - Comparison of annual expenses per patient

As expected, we observed significant differences in healthcare resource utilization between rare disease patients and those with other conditions (Figure 3), with higher rates of consultations, emergency services, tests, therapies, and hospitalizations ($p<0.001$).

Figure 3 – Healthcare resource utilization rates, average per patient/year

HCRU	RDs	CCs	*p value*
Visits	5.2	3.8	<0.001
Tests	51.6	32.5	<0.001
Therapies	11.7	4.4	<0.001
Emergency services	46.2%	37.7%	<0.001
Hospitalizations	26.9%	13.0%	<0.001

CONCLUSIONS

Despite the challenges in quantifying rare diseases within healthcare systems using ICD coding search criteria—likely leading to an underestimation of their true impact—in our study, using a custom list of conditions, we observed that rare diseases collectively affect a significant number of people and incur very high costs, approximately three to four times greater than those of the common conditions group.

This is primarily due to prolonged diagnostic journeys that can lead to progressive, irreversible, and costly complications, creating an extraordinary financial and organizational challenge for payers.

With innovative real-world studies of this nature, we believe it is possible to optimize healthcare resource utilization and achieve better clinical outcomes for patients, expanding access to the best available care. In doing so, we hope to support the healthcare system in addressing rare diseases.

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