

# REAL-WORLD HEALTHCARE RESOURCE UTILIZATION AND RELATED COSTS ASSOCIATED WITH ULTRARARE DISEASES IN THE BRAZILIAN PRIVATE MARKET

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## BACKGROUND

There is no internationally standardized concept regarding which diseases are rare or ultrarare, but usually an ultrarare disease (UD) is defined as a debilitating or life-threatening condition with an incidence of no more than 1 case per 50,000 inhabitants. Currently, although the prevalence of each disease individually is low, there are millions of people in the world who suffer from one of these diseases. Most ultrarare diseases, around 95%, do not have a defined treatment protocol and require specialized rehabilitation services.

## OBJECTIVES

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

## 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. From this list, we identify conditions with prevalence less than or equal to 1 case for every 50,000 in our population resulting in a database of 289 ICD-10 list of UD. For comparative purposes, we defined another group of individuals with common conditions (CCs), not included neither in this list nor on own list linked as of rare diseases primarily provided by Orphanet.

**1 case in 50,000** database of **289 ICD-10** of UD list

This retrospective cohort study included all beneficiaries who had a record in the health plan's administrative database with one of the 289 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  $p < 0.05$ .

## REFERENCES

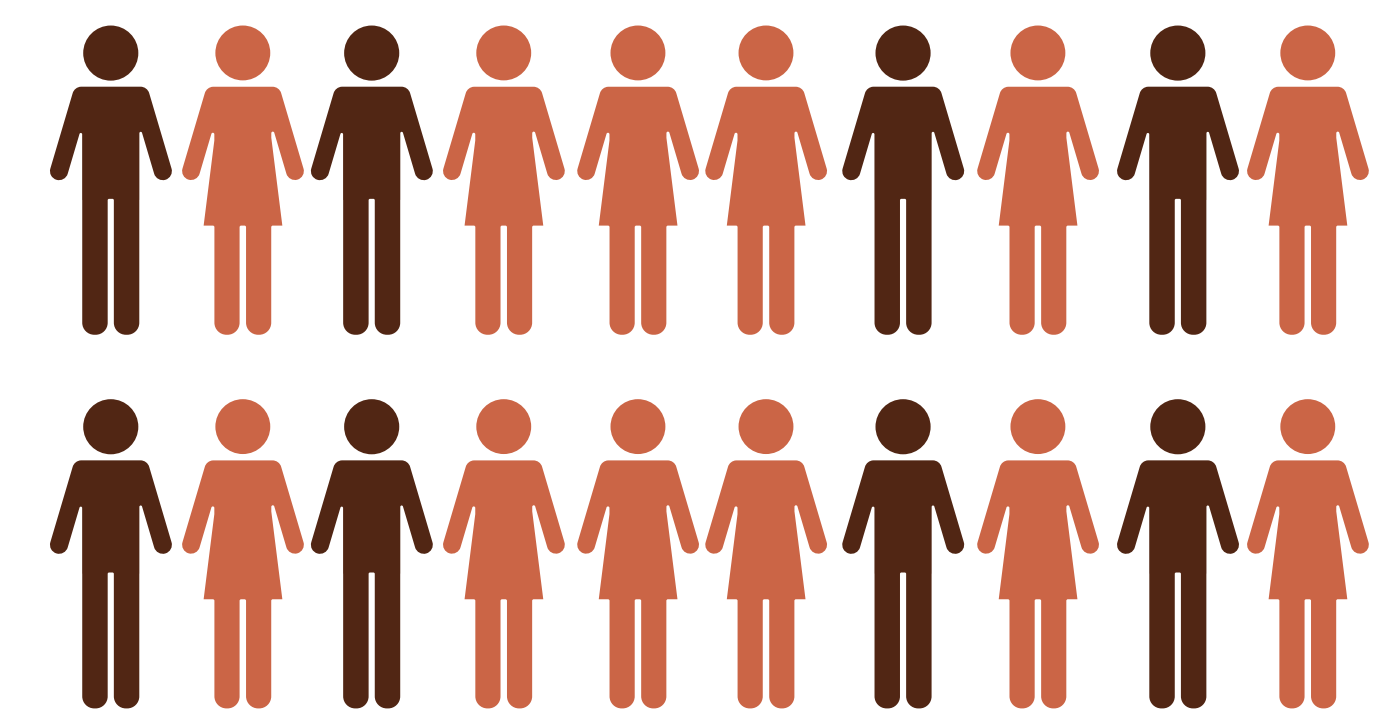
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## RESULTS

A total of 60,012 individuals (average age 53 years) were analyzed. When combined, UD collectively affect 0.5% ( $n=283$ , 61 years old, female 61.1%).

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Regarding healthcare expenses over the 5-year study period, these totaled slightly over \$295 million in health plan costs, with 2.7% (\$8 million) attributed to UD and \$287 million to common conditions, despite ultrarare disease patients representing only a small percentage of the total population.

The study shows significant differences in HCRU between UD and CCs (Figure 1) with a higher utilization for visits, emergency services, tests, therapies and hospitalizations when compared to CCs ( $p < 0.0001$ ), and nearly quintuple (Figure 2) the average total charges per patient (\$8,646) compared to CCs (\$1,960) ( $p < 0.0001$ ).

Figure 1 – Healthcare resource utilization rates, average per patient/year

HCRU	UDs	CCs	<i>p value</i>
Visits	5.4	3.8	<0.001
Tests	57.0	32.5	<0.001
Therapies	13.2	4.4	<0.001
Emergency services	51.7%	37.7%	<0.001
Hospitalizations	32.9%	13.0%	<0.001

Figure 2 - Comparison of annual expenses per patient

UDs  
\$8,646

VS

CCs  
\$1,960

## CONCLUSIONS

In our study, we observed that UD collectively affects a small number of people, but incurs very high costs, approximately five 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. This paper will increase the knowledge of UD and help health managers to improve national policy on ultrarare diseases in Brazilian private market.