

# EPIDEMIOLOGY, HEALTHCARE RESOURCE CONSUMPTION AND RELATED COSTS FOR HYPOPHOSPHATEMIC VITAMIN D RESISTANT RICKETS PATIENTS IN AN ITALIAN REGION

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## OBJECTIVE

Hypophosphatemic vitamin D resistant rickets is a rare hereditary disorder characterized by an increased urinary phosphate excretion that interferes with the process of bone mineralisation. Clinical features comprise growth retardation and bones deformities of the limbs -especially lower limbs- associated with neuromuscular pains.

The present study aimed to perform an epidemiologic analysis of patients with hypophosphatemic vitamin D resistant rickets in a real-world setting in an Italian Region, and also to evaluate in the study population the healthcare resource consumption and related costs for the Italian National Health System (INHS) in terms of drug treatments, specialist visits and hospitalizations.

## METHODS

- An observational retrospective cohort analysis was conducted based on administrative databases of an Italian Region (Veneto Region).
- All patients with the exemption code for hypophosphatemic vitamin D resistant rickets RC0170 between 01/01/2010 and 31/12/2016 (inclusion period) were included. Index date was defined as the date in which the patient presented the exemption code during inclusion period. Patients who were transferred to another Region during the study period were excluded from the analysis.
- All patients included in the analysis were characterized one year before index date and followed-up from index date to the end of study (*follow-up* period).
- Presence of comorbidities was measured using the Charlson Comorbidity Index (CCI).
- Healthcare resource consumption and related mean annual costs per patient were estimated during the last available 3 years of follow-up. The cost analysis was conducted with the perspective of the INHS.

## RESULTS

A total of 57 patients affected by hypophosphatemic vitamin D resistant rickets were included and mean age  $\pm$  Standard Deviation (SD) was  $31.4 \pm 24.3$  years. 45.6% of included patients were male. In our analysis, prevalence rate of hypophosphatemic vitamin D resistant rickets was estimated to be 1.2 cases per 100,000 individuals. Demographic and clinical characteristics of study population (N= 57) are reported in Table 1.

**Table 1. Characteristics of included patients.**

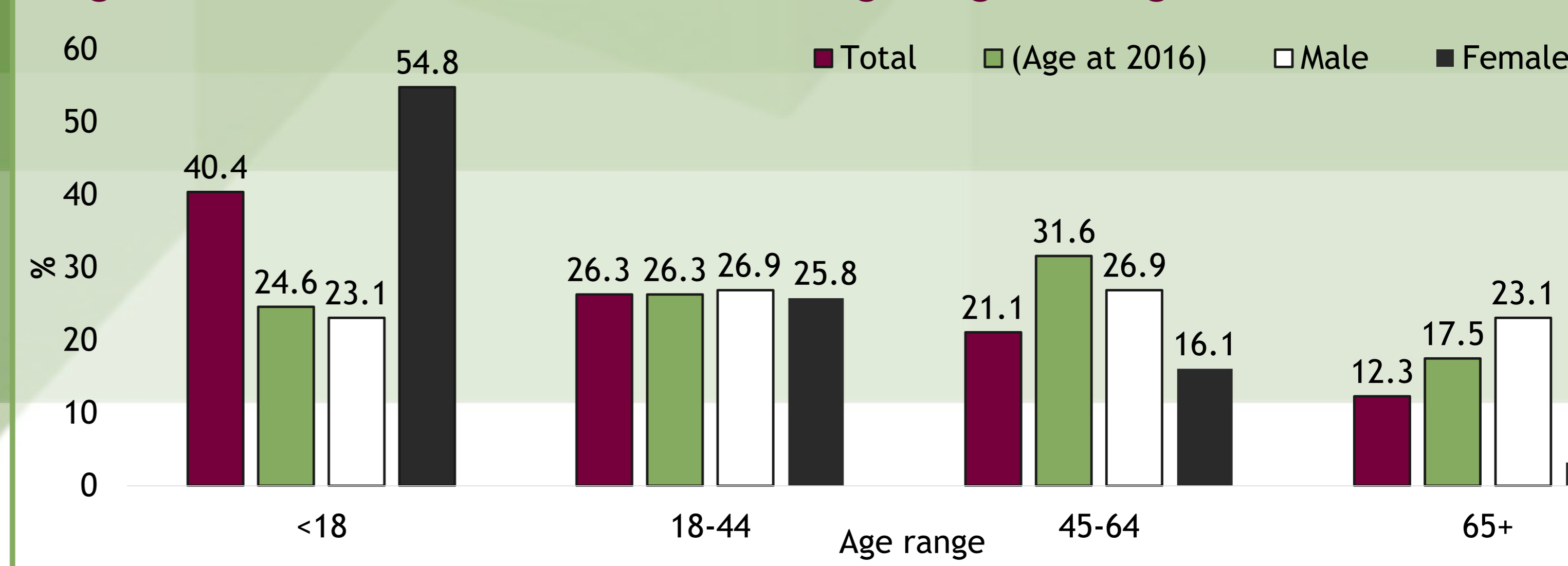
	Included patients N=57
Prevalence per 100,000* (%)	1.2
Age, mean $\pm$ SD at 31/12/2016	37.9 $\pm$ 22.9
Age, mean $\pm$ SD at diagnosis *	31.4 $\pm$ 24.3
Male, n (%)	26 (45.6)
Charlson Comorbidity Index, mean (SD)	0.39 $\pm$ 0.59

Note. \*Prevalence rate was calculated considering as proxy of diagnosis the presence of the exemption code for hypophosphatemic vitamin D resistant rickets (code: RC0170) and the possible concomitant presence of a discharge diagnosis of hypophosphatemia (ICD-9-CM code: 275.3).

## RESULTS (CONTINUED)

Figure 1 shows the distribution of patients according to age range at diagnosis. At the time of the analysis (age at 2016), 24.6% (n=14) of included patients were under age 18 and their mean age  $\pm$  SD was  $6.4 \pm 5.4$  years.

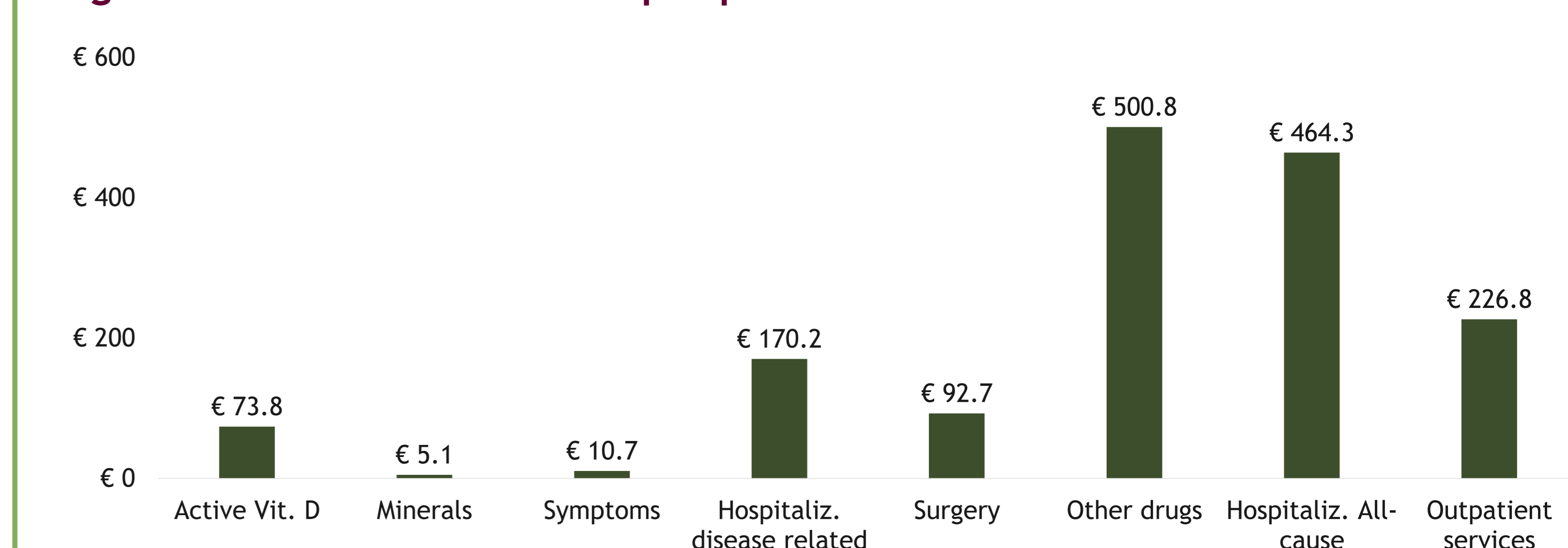
**Figure 1. Patients distribution according to age at diagnosis.**



Note. Date of exemption code was used as proxy of diagnosis date.

Considering healthcare resource consumption, during the last 3 years of follow-up, 7% of patients underwent orthopedic surgery, while 28.1% were hospitalized for all-cause. The mean annual healthcare costs per patient included in the study was € 1,544.4, with a maximum annual cost of € 11,553.7. Annual costs related to each resource consumption expressed as mean and median with interquartile ranges is shown in Figure 2.

**Figure 2. Overall annual costs per patient.**

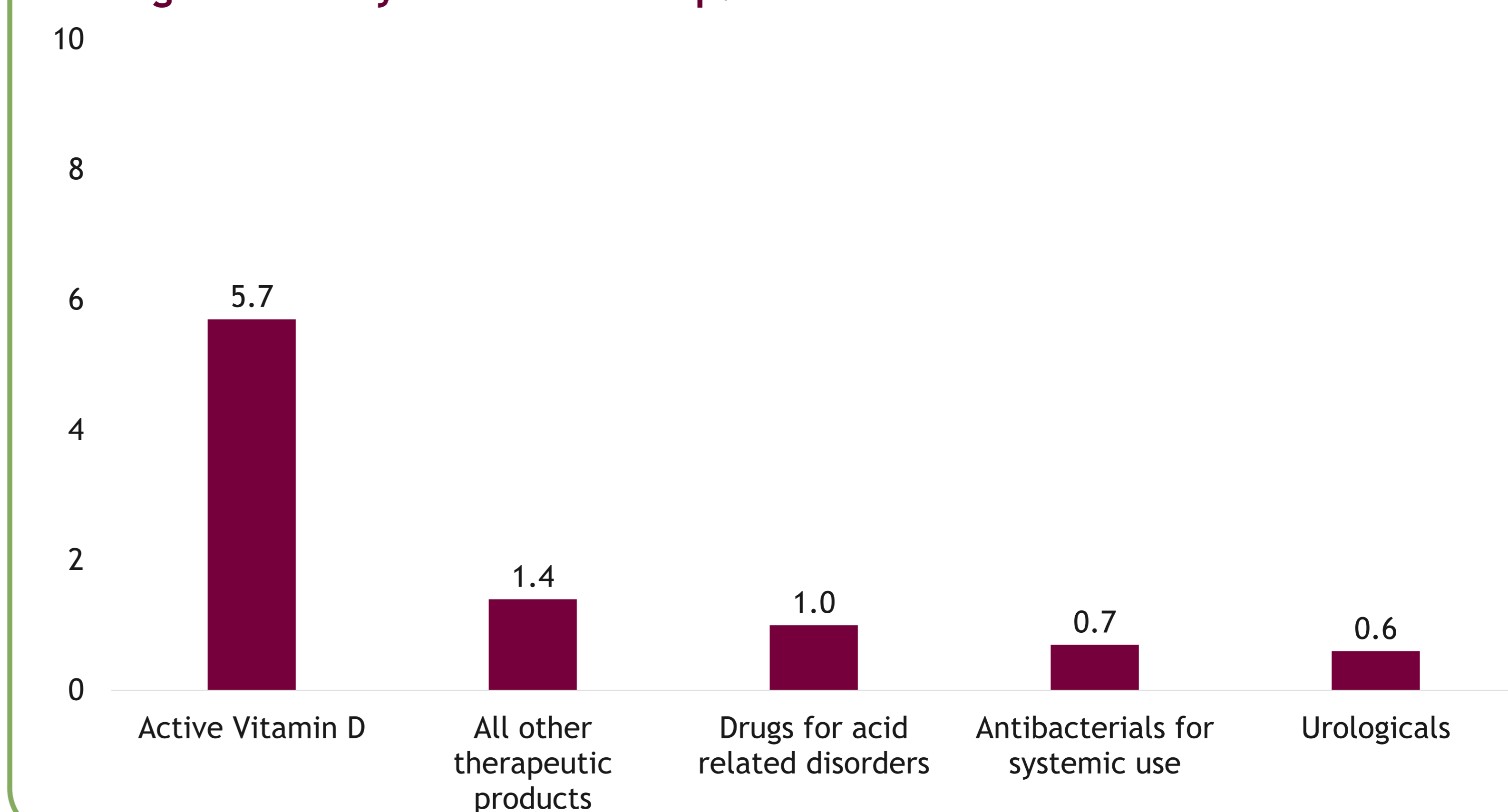


	Active Vitamin D	Minerals*	Symptoms	Hospitalization disease related	Surgery	Other drugs	Hospitalization all-cause	Outpatient services
Minimum	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0
Quartile 1	11.7	0.0	0.0	0.0	0.0	64.5	0.0	32.1
Median	40.6	0.0	0.0	0.0	0.0	203.3	0.0	98.3
Quartile 3	115.2	5.7	0.0	0.0	0.0	368.1	461.7	222.2
Maximum	326.4	210.7	73.7	9,699.0	2,288.3	7,441.0	7,618.9	2,140.4

Note. \*identified by ATC code: A12.

As for drug treatment, over the last 3 years of follow-up active Vitamin D analogs were the drugs mainly prescribed, with a mean annual prescription number of 5.7 per patient. Figure 3 reports the mean annual prescription number of the drugs most frequently used during the last 3 years of follow-up.

**Figure 3. Mean annual prescription number of drugs most frequently used during the last 3 years of follow-up.**



## CONCLUSIONS

Prevalence calculated in our analysis was lower than the expected prevalence reported in literature (1:20,000)<sup>1</sup>. This discrepancy could be explained considering the methodology applied, since we included in the analysis only patients presenting the diagnosis of hypophosphatemic vitamin D resistant rickets identified by exemption code. Moreover, the low prevalence herein reported could suggest a problem of underdiagnosis, which is commonly observed among rare diseases.

## References

1. Orphanet: X linked hypophosphatemia. [https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Expert=89936&lng=EN](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=89936&lng=EN). Accessed October 5, 2019.