

Objectives

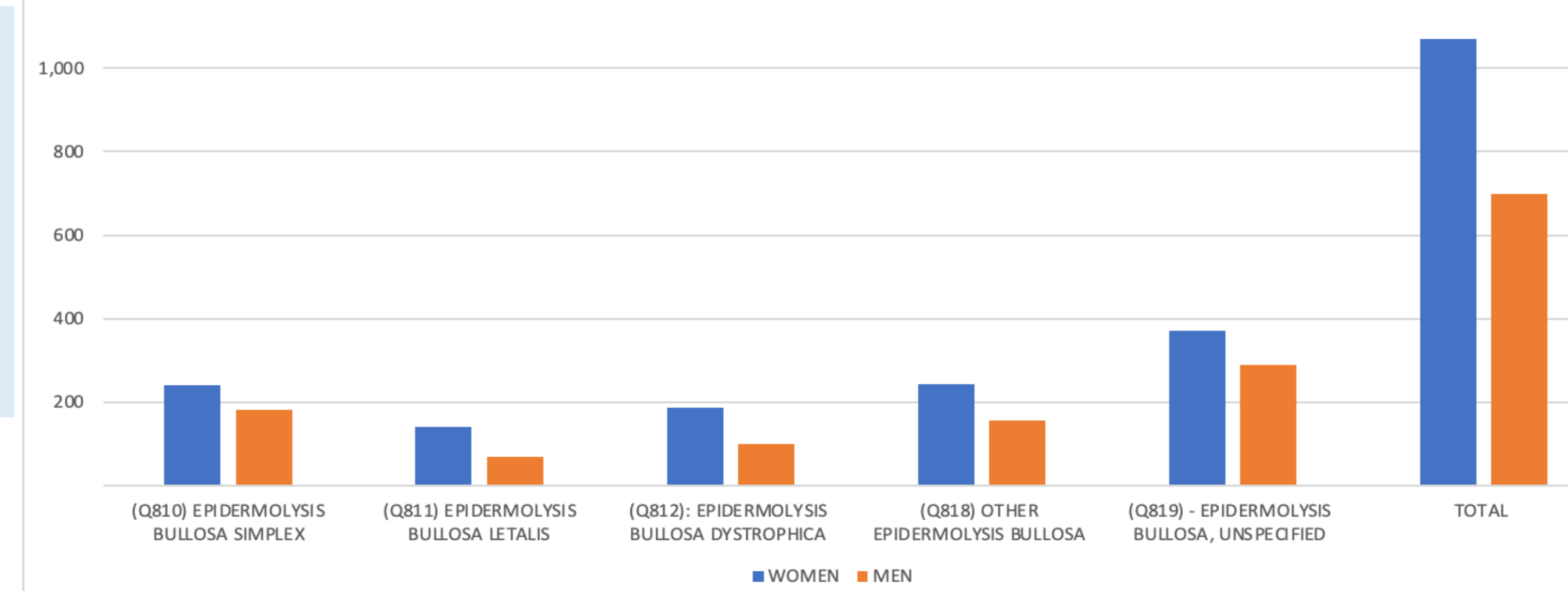
Epidermolysis bullosa (EB) is a heterogeneous group of hereditary diseases characterized by varying degrees of skin mucosa fragility caused by autosomal recessive mutations that affect skin structural proteins. Its prevalence in Colombia and most latin American countries is unknown. Under the leadership of the Ministry of health a rare disease registry has been developed, we used it to study age, gender and geographic distribution of the disease.

Methods

We accessed the RIPS database, years 2018-2022, using ICD-10 codes for EB simplex; letalis; dystrophica, unspecified and others, organized by gender, age and place of residence. The prevalence was estimated per million inhabitants. For geographic distribution Colombia was divided in 5 regions (Amazon, Andes, Caribbean, Orinoco and Pacific).

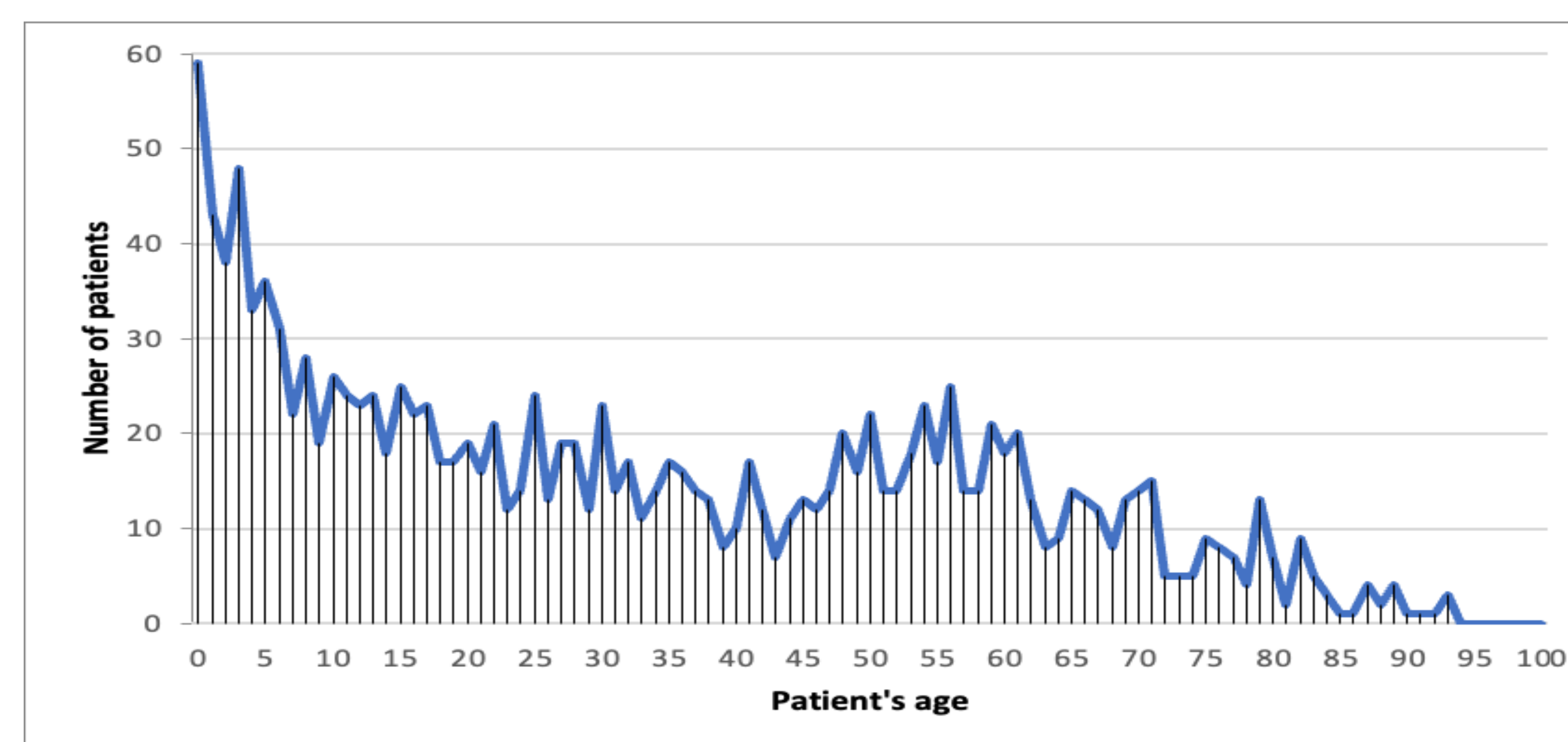
Results

1774 patients were registered with the diagnosis of EB. 1070 were females (60,3%) for female:male ratio of 1.53. The mean age was 26.9 years. The prevalence for all types of diagnosis in Colombia was 34.1 per one million live births .



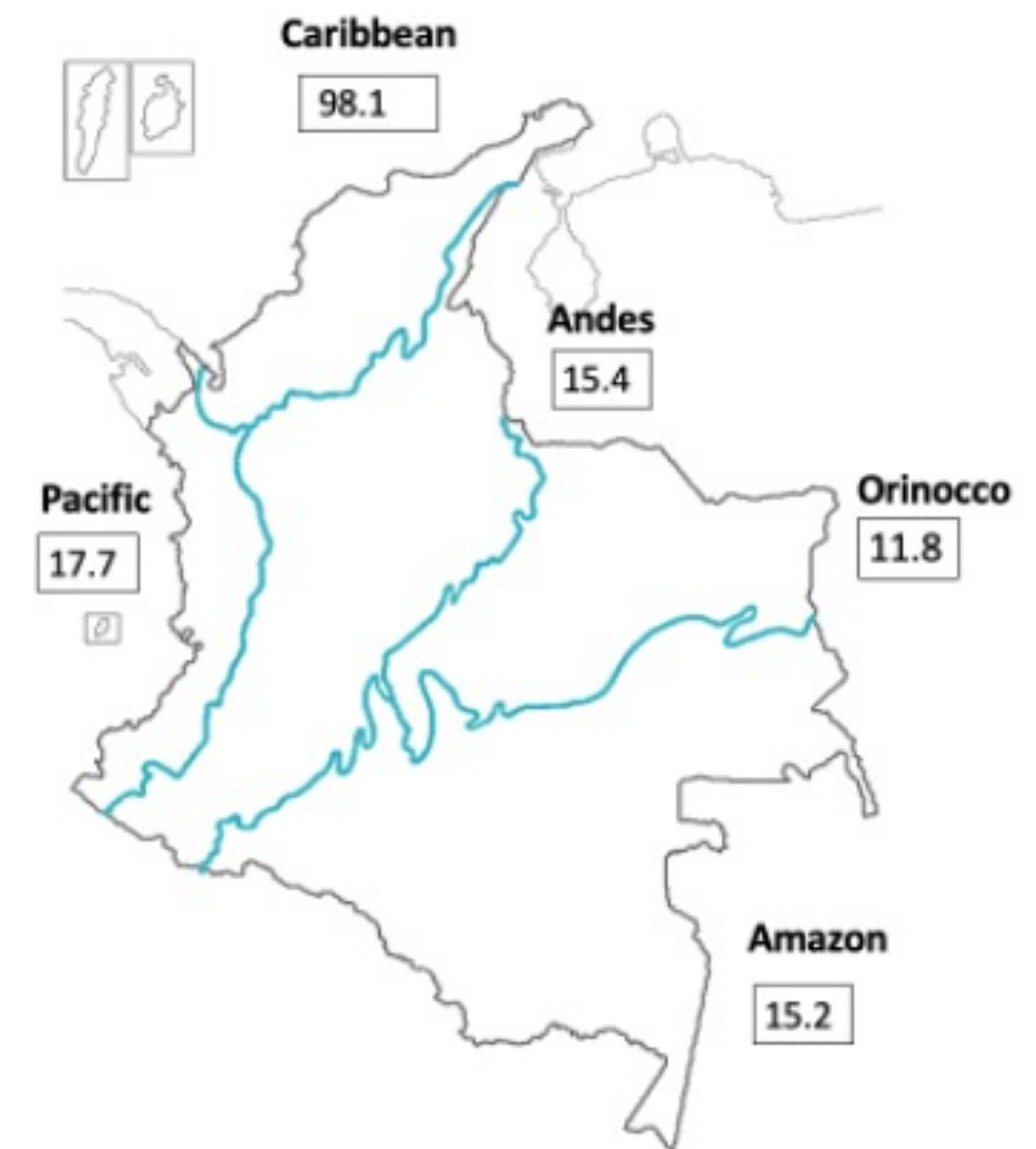
Source: RIPS

Figure 1. Number of cases according to gender and ICD 10 code



Source: RIPS

Figure 2. Age of patients with of Epidermolysis bullosa in Colombia.



Source: RIPS

Figure 3. Prevalence per million in each region of Colombia.

Overall prevalence per million in each region was: Caribbean 98.1, Pacific 17.7, Andes 15.4, Amazon 15.2 and Orinoco 11.8.

- The Caribbean Region had the largest number of patients 1185 (66.8% of the total), and also the highest prevalence rates in the departments of Córdoba (175.0), Atlántico (124.8), and Sucre (108.1).
- EB letalis, the most severe form of the disease , represented 16.5% of all cases in the Caribbean but only 2.2% in the rest of the country.

Conclusions

EB in Colombia is a hereditary disease that is part of the orphan or rare diseases that has a greater prevalence in women, with a majority of patients diagnosed in the first 5 years of age. The Caribbean region may have one of the highest prevalence rates and most severe disease profiles. Genetic and other epidemiological studies are needed to better explain the reasons for this high concentration of cases.