In recent years it is increasingly normal to hear information about rare diseases, where the lack of knowledge about these rare diseases causes a high number of deaths without being able to treat them due to not having an effective medicine. To be called a rare disease, it has to affect at least 5 patients per 10,000 and of these we know that there are at least 6,000 diseases. In a portion of 80% rare diseases affect at least one patient in a million, making it very difficult to diagnose the patient even for the most experienced doctors.

New methods of machine learning are transforming medicine and health care information is very useful for the diagnosis and treatment of these diseases. This study tries to investigate how machine learning through types of algorithms and input data can be useful in diagnosis, prognosis or treatment.

The procedural method of the study was as follows, a complex search was made with generic terms on the official PubMed page, where they have found a result of 381 names of individual diseases in the last 10 years. Terms such as the location of the different types of trials were added to obtain a greater source of information. To proceed with the analysis, the studies have been arranged according to different types of dimensions.
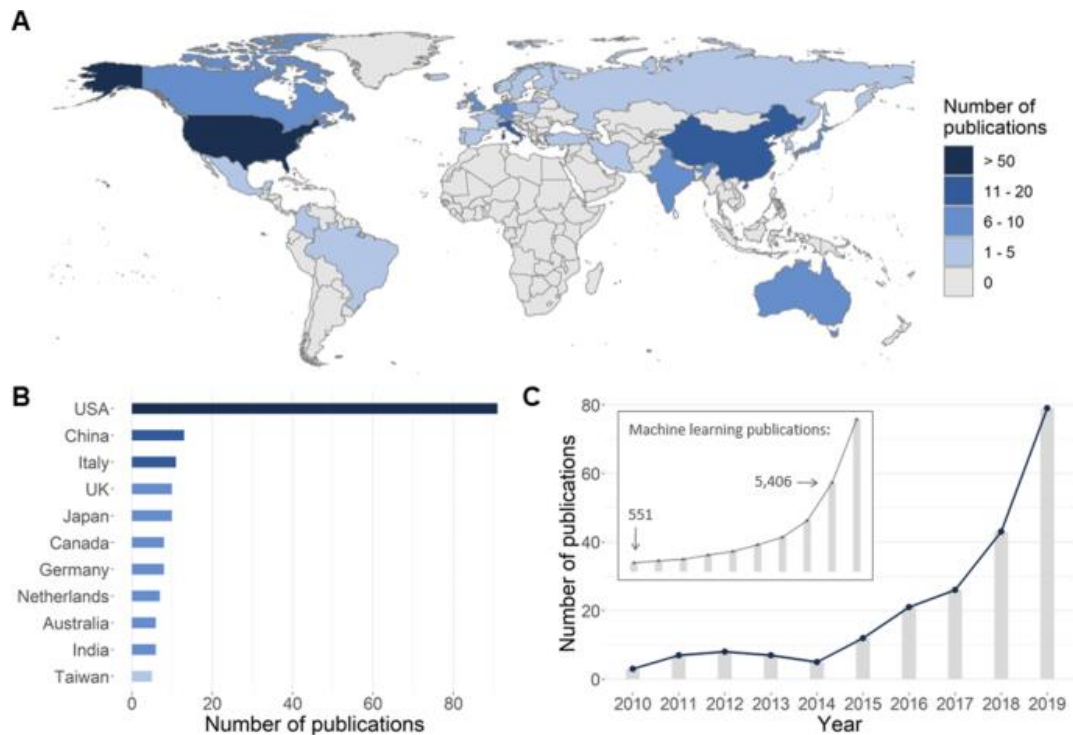
Figure 1. It shows us the number of rare disease studies in different locations.

The analyzes obtained showed us that there are a total of 211 studies in a total of 32 countries that studied 74 rare diseases. It has also been seen that diseases with a higher prevalence had a higher frequency in clinical studies, although with the exception of some rare neurological diseases, systemic or rare rheumatological diseases. Learning as a whole (36%), support vector machines (32.2%) and artificial neural networks (31.8) has been the most applied algorithm in studies and, above all, it should be noted that almost 40.8% of machine learning studies for diagnosis and prognosis in 38.40%. With a too low percentage, around 4.7% of machine learning studies have been dedicated to the treatment of these rare diseases.
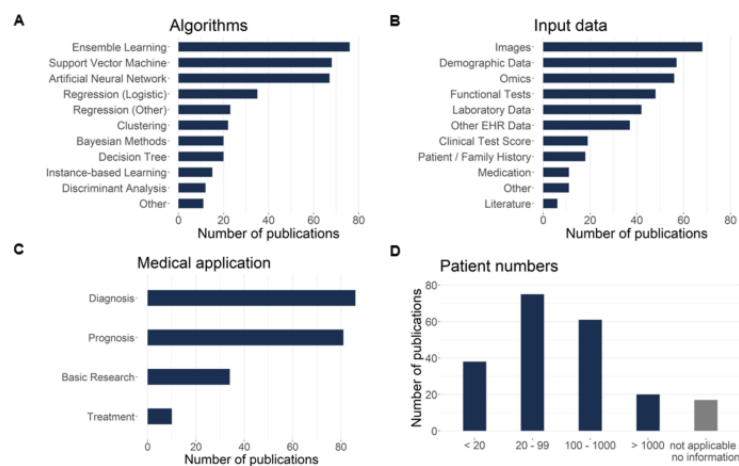

Figure 2. Percentage of studies dedicated to different types of approaches and their algorithms.

We can conclude that this new learning model is increasingly being used to help with these rare diseases, where we could obtain greater knowledge of their origin, spread and especially in the treatment of these.

- (1,2) : Schaefer J, Lehne M, Schepers J, Prasser F, Thun S. The use of machine learning in rare diseases: a scoping review. Orphanet J Rare Dis. 2020 Jun 9;15(1):145. doi: 10.1186/s13023-020-01424-6. PMID: 32517778; PMCID: PMC7285453.