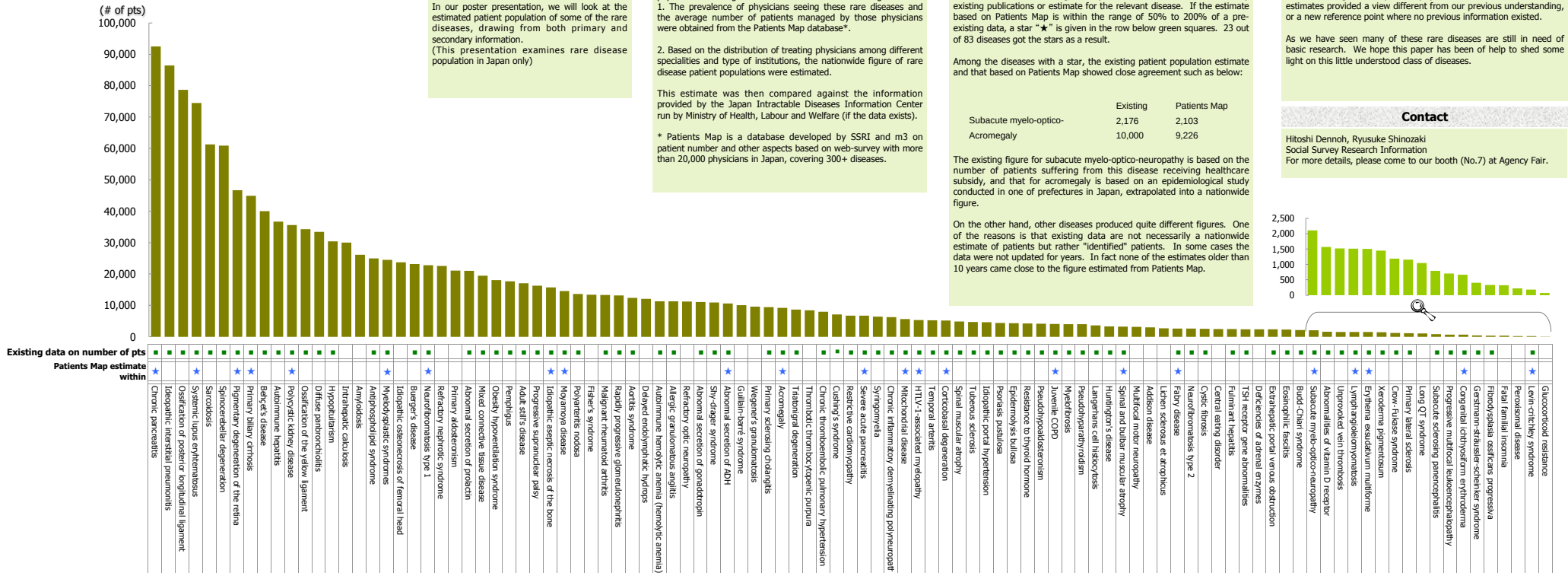
How Common are Rare Diseases?

Social Survey Research Information

Estimated Rare Disease Population in Japan



Introduction

Rare diseases are attracting much attention of pharmaceutical companies as the next frontier in pharmaceutical research and development. However, due to its very nature of being rare, these diseases have been difficult to understand.

In our poster presentation, we will look at the estimated patient population of some of the rare diseases, drawing from both primary and secondary information.
(This presentation examines rare disease population in Japan only)

Methodology

In Japan, 130 diseases are designated by the Japanese government as rare and intractable diseases, research and development for which are encouraged and subsidised.

For the purposes of this poster presentation, we estimated the population suffering from these diseases in the following manner:
1. The prevalence of physicians seeing these rare diseases and the average number of patients managed by those physicians were obtained from the Patients Map database*.

2. Based on the distribution of treating physicians among different specialities and type of institutions, the nationwide figure of rare disease patient populations were estimated.

This estimate was then compared against the information provided by the Japan Intractable Diseases Information Center run by Ministry of Health, Labour and Welfare (if the data exists).

* Patients Map is a database developed by SSRI and m3 on patient number and other aspects based on web-survey with more than 20,000 physicians in Japan, covering 300+ diseases.

Results and Discussions

The chart below shows the estimated patient population of the rare diseases based on Patients Map. The estimated numbers of patients ranged from over 90,000 to under one hundred. 90% of the diseases have more than 1,000 patients and 43% have more than 10,000.

The row below that with green square shows whether there are any existing publications or estimate for the relevant disease. If the estimate based on Patients Map is within the range of 50% to 200% of a pre-existing data, a star "★" is given in the row below green squares. 23 out of 83 diseases got the stars as a result.

Among the diseases with a star, the existing patient population estimate and that based on Patients Map showed close agreement such as below:

	Existing	Patients Map
Subacute myelo-optico-	2,176	2,103
Acromegaly	10,000	9,226

The existing figure for subacute myelo-optico-neuropathy is based on the number of patients suffering from this disease receiving healthcare subsidy, and that for acromegaly is based on an epidemiological study conducted in one of prefectures in Japan, extrapolated into a nationwide figure.

On the other hand, other diseases produced quite different figures. One of the reasons is that existing data are not necessarily a nationwide estimate of patients but rather "identified" patients. In some cases the data were not updated for years. In fact none of the estimates older than 10 years came close to the figure estimated from Patients Map.

Conclusions

We estimated the number of patients suffering from rare diseases in Japan by using the results of a large-scale web survey, and compared it against the existing figure on patient population.

In some areas our new estimates corroborated the existing understanding of the patient population. In other areas, our estimates provided a view different from our previous understanding, or a new reference point where no previous information existed.

As we have seen many of these rare diseases are still in need of basic research. We hope this paper has been of help to shed some light on this little understood class of diseases.

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