

1. INTRODUCTION

In Japan, access to treatment for rare diseases is particularly limited due to drug lag and loss, with nearly half of the unavailable or withdrawn drugs being those intended for rare conditions¹. Consequently, many patients have no choice but to rely on symptomatic care, which often fails to sufficiently stabilize their condition².

Burden of rare diseases

The burden of rare diseases extends beyond clinical challenges. A recent retrospective study using medical and insurance records suggests that health care costs for individuals with rare diseases are three to five times higher than those without such conditions³. They also highlighted the medical expenditures for rare diseases may have been underestimated.

In addition to financial strain, patients with rare diseases frequently experience significant physical and psychological burdens^{2,4}, emphasizing the need for comprehensive health care and policy support.

Definitions of rare, intractable disease in Japan

In Japan, rare diseases and intractable diseases are defined by specific legal frameworks. Rare disease is a condition that affects fewer than 50,000 people in Japan⁵. An intractable disease is one with an unclear pathogenesis, no established treatment, rarity, and a need for long-term care⁶. To be selected as a designated intractable disease, the condition must be objectively diagnosable and affect ~0.1% or less of the population.

Act on Medical Care for Patients with Intractable Diseases

Under the system established in 2015, patients diagnosed with a designated intractable disease may be eligible for medical expense subsidies under the “Act on Medical Care for Patients with Intractable Diseases” if they meet either of the following criteria:

- 1. The severity of the condition meets or exceeds a specified threshold based on the severity classification
- 2. Although the condition does not meet the severity classification criteria, the total monthly medical expenses exceed JPY 33,330 for at least three months within a year

2. PURPOSE

This study aims to evaluate the current health care policy initiatives that support patients with rare and intractable diseases in Japan.

3. METHODS

A targeted review of government websites and gray literature was conducted to identify policy measures aimed at alleviating the economic burden associated with rare and intractable diseases in Japan. All searches were conducted in Japanese, focusing on publicly available information from key government sources, including the Ministry of Health, Labour and Welfare (MHLW), Ministry of Finance, the Child Chronic Disease Information Center⁶, and the Intractable Disease Information Center⁷.

4. RESULTS

Trends among patients with designated intractable diseases

- ❖ Designated intractable diseases were first recognized in January 2015, with 110 conditions initially eligible for medical subsidies. The coverage has gradually expanded, with additional conditions added in July 2015, April 2017, April 2018, July 2019, November 2021, and April 2024. In April 2025, seven more diseases were included, bringing the total to 348 conditions eligible for medical expense subsidies.
- ❖ The number of individuals eligible for medical expense subsidies has slightly increased over the past decade (Figure 1).

- ❖ With the enforcement of the Intractable Disease Law in 2015, all patients had to meet new diagnostic and severity criteria⁸. Some became ineligible or did not reapply, leading to a drop in the number of patients receiving financial benefits when the transition period ended in 2017.

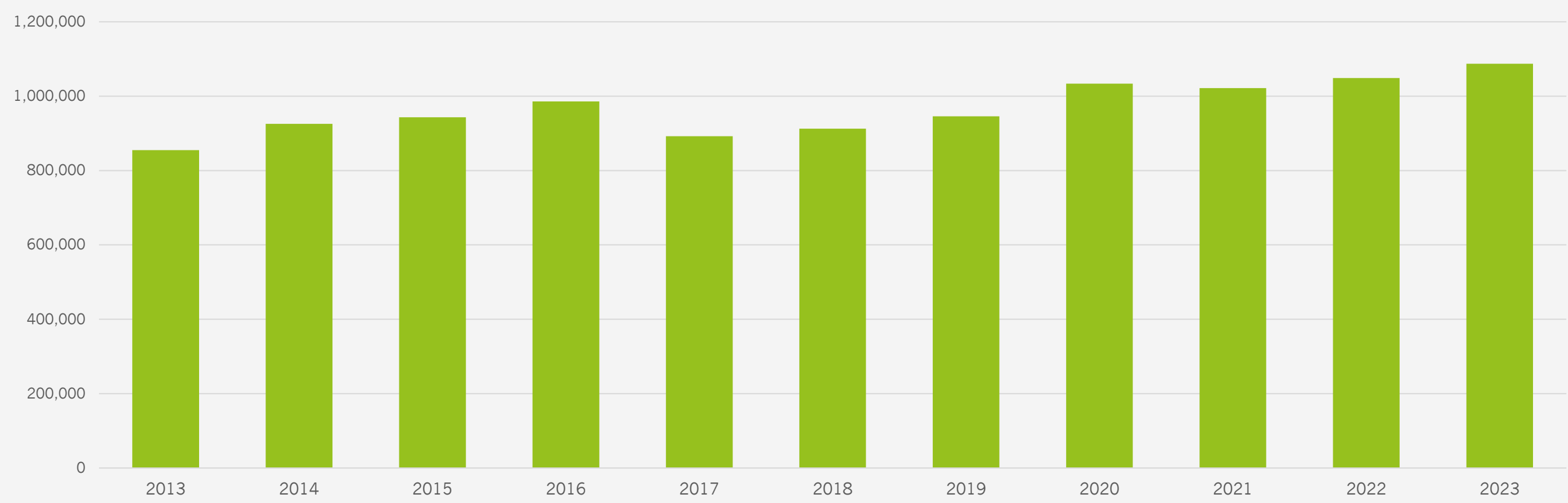


Figure 1. Trends in the number of individuals eligible for medical expense subsidies for designated intractable diseases

- ❖ In 2023, autoimmune and inflammatory diseases were among the most common (Table 1), and patients aged 60 and older accounted for the greatest share (Figure 2).

Table 1. Patient counts and total share for the five most common diseases (2023)

Disease name	N	%
Parkinson's disease	147,481	13.6%
Ulcerative colitis	146,702	13.5%
Systemic lupus erythematosus	66,297	6.1%
Crohn's disease	52,108	4.8%
Ossification of the posterior longitudinal ligament	31,733	2.9%

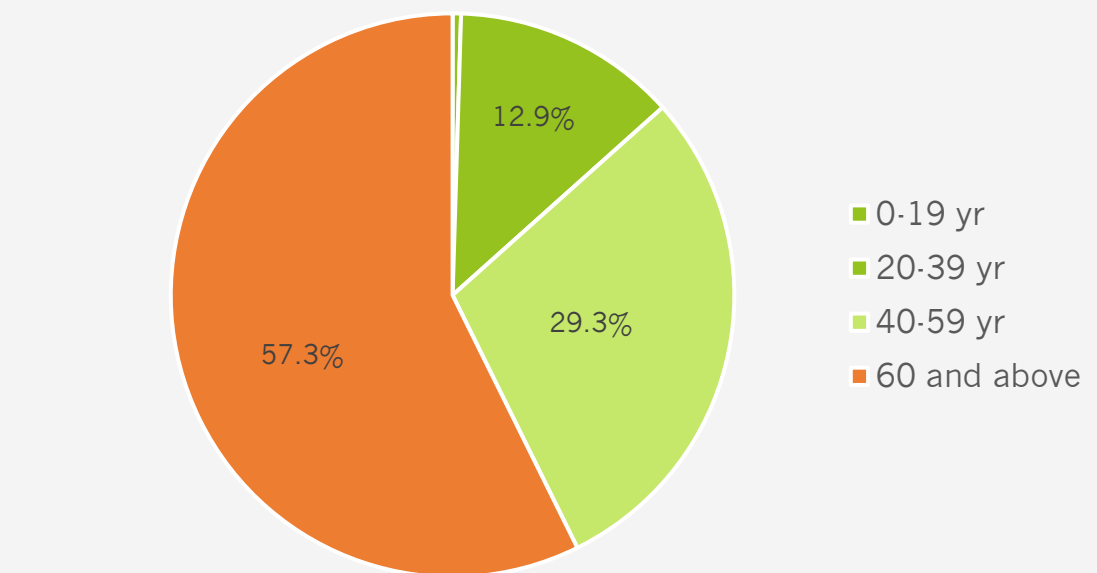


Figure 2. Age distribution of patient (2023)

Trends among paediatric patients with designated chronic diseases

- ❖ Medical expenses for children with designated paediatric chronic diseases are subsidized. To qualify, the disease must meet the following criteria: it is a chronic condition that persists over time, it poses a long-term threat to life, its symptoms or treatment significantly impair quality of life over an extended period, and it requires prolonged and costly medical care.
- ❖ In April 2025, 13 more diseases were included, bringing the total number of conditions eligible for medical expense subsidies to 801.
- ❖ With support from the MHLW, the development of a database for children with designated paediatric chronic diseases has been underway since 2015. According to the database, approximately 100,000 patients are currently registered (Figure 3). As the database is still under development, the actual number of patients is likely higher. A large proportion of registered patients are those with endocrine diseases (Table 2).

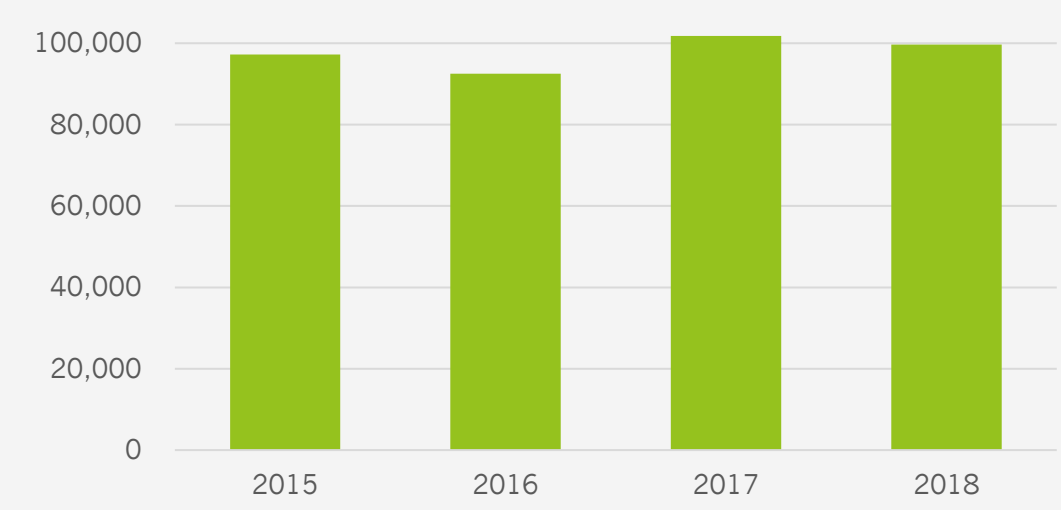


Figure 3. Trend in the number of registered patients

Table 2. Patient counts and total share for the five most common diseases (2018)

Disease name	N	%
Growth hormone deficiency-related short stature	9,142	9.2%
Type 1 diabetes mellitus	5,140	5.2%
Congenital hypothyroidism	3,523	3.5%
Precursor B-cell acute lymphoblastic leukemia	3,328	3.3%
West syndrome	2,943	3.0%

High-cost medical expense benefit

- ❖ The high-cost medical expense benefit is a program in which, if the amount paid at medical institutions or pharmacies exceeds a monthly cap, the excess amount is reimbursed. The monthly cap varies depending on age and annual income.

- ❖ This system helps reduce out-of-pocket medical expenses to some extent, not only for patients with intractable diseases.
- ❖ This system was revised on December 25, 2024, to address the increase in medical expenses and adjust the balance of burden across generations. These updates include an increase in the monthly copayment cap (effective August 2025; Table 3) and an expansion of income categories (effective August 2026) — However, in response to opposition from political parties and patient advocacy groups it has been put on hold (on March 3, 2025).

Table 3. Changes to the monthly cap under the high-cost medical expense benefit system (younger than 70 years of age)

Income level	Monthly cap amount (~July 2025) JPY	Monthly cap amount (Augst 2025~) JPY
Annual income ≥ 11.6 million	252,600 + (Medical expenditure - 842,000) x 1%	290,400 + 1%* (increase of 37,800)
7.7 million ≤ Annual income < 11.6 million	167,400 + (Medical expenditure - 558,000) x 1%	188,400 + 1%* (increase of 21,000)
3.7 million ≤ Annual income < 7.7 million	80,100 + (Medical expenditure - 267,000) x 1%	88,200 + 1%* (increase of 8,100)
1.56 million ≤ Annual income < 3.7 million	57,600	60,600 (increase of 3,000)
Residential tax exemption individual	35,400	36,300 (increase of 900)

*+1% refers to requiring patients to pay 1% out-of-pocket for medical expenses that exceed the standard copayment amount.

Future revisions to Japan’s medical expense subsidy framework

- ❖ Given the limitations of national financial resources, there is an ongoing discussion about revising the list of diseases eligible for medical expense subsidies under the designated intractable disease program.
- ❖ The criteria for designation are being reviewed, and the implementation of revised eligibility requirements for individual diseases is being considered from 2026 onward.
- ❖ If treatments emerge or prognoses improve, such diseases may be delisted to redirect support to those with no effective treatment or poor outcomes.
- ❖ The government intends to review the overall framework of the high-cost medical expense benefit system beyond August 2026 and aims to reach a conclusion by 2025 autumn.

5. CONCLUSION

Several policies are in place in Japan to help alleviate the financial burden on patients with rare and intractable diseases. The expansion in the number of conditions eligible for pediatric chronic disease and rare disease subsidies is a positive development. However, due to ongoing concerns about securing sufficient funding, existing support schemes—including the high-cost medical expenditure subsidy—are under review, and patients may face higher out-of-pocket costs in the future.

References

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