Intractable & Rare Diseases 2025

10 Years After the Passing of the Intractable Diseases Act, Toward a Co-created Future

HGPI Health and Global Policy Institute

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Health and Global Policy Institute (HGPI) Intractable & Rare Diseases Project Discussion Paper

Considerations for Future Policy on Intractable & Rare Diseases

Reflections from the Perspective of Patients and the Public 10 Years After the Passing of the Intractable Diseases Act

Background to this discussion paper

Since its establishment as a non-profit, independent, non-partisan health policy think-tank in 2004, Health and Global Policy Institute (HGPI) has worked to present society with policy options in line with our mission to achieve citizen-centered health policy. Although the Intractable & Rare Diseases Project is a new project launched in FY2024, the theme is familiar for HGPI as we have long been engaged in projects that address various chronic diseases. Intractable diseases often have unclear mechanisms of occurrence, and are rare diseases that still lack definitive treatments. Moreover, these diseases often require long-term medical care, frequently placing significant physical and emotional burdens on the patients and their families.

Japan currently designates 341 conditions as "intractable diseases," which are said to affect over one million patients throughout the country. Ever since the compilation of the "Outline of Intractable Disease Measures" in 1972, the Government of Japan has been advancing measures for intractable and rare diseases with a focus on three core areas: promoting R&D, improving the healthcare provision system, and supporting patients (mainly through the subsidization of care). The year 1998 saw the establishment of "Liaison Councils for Intractable Disease Treatment" and "Intractable Disease Support Centers" in each prefecture. Concurrently, efforts were launched to provide various welfare programs, including initiatives to improve patients' living environments and provide employment support. The Long-Term Care Insurance System, established in 2000 to provide support to the elderly, also accommodates cases in which care is required due to intractable diseases. In 2014, the Act on Medical Care for Patients with Intractable Diseases (Intractable Diseases Act) was enacted, setting a direction not only for promoting healthcare, but also for improving the social environment. In FY2025, the designation criteria for intractable diseases will be updated. In this way, systems continue to evolve to reflect advancements in medicine.

However, measures for intractable and rare diseases face a number of lingering issues. For the healthcare provision system, these include information disparities, in which people do not receive enough of the information they need; long diagnostic delays (diagnosis lag); regional healthcare disparities; and issues related to transitional care. There are also a number of actions that should be taken to promote innovation. For example, we hope to see the establishment of a data aggregation system to further basic research on each disease, as well as efforts to address issues such as drug lag, where treatments approved overseas are not yet available in Japan, and drug loss, where certain treatments have not been developed at all—particularly in the context of orphan drugs (medications for rare diseases). We also hope to see more patient and public involvement (PPI), as well as medical digital transformation (DX) to ensure a smooth flow from diagnosis to treatment.

As part of the HGPI Intractable & Rare Diseases Project, we held the Intractable Disease Day Symposium on May 23, 2024 (Intractable Disease Day), the 10th anniversary of the enactment of the Intractable Diseases Act. As a discussion paper outlining future policy directions as of the present point in time, this document compiles points for consideration based on the discussions held at the symposium, subsequent desktop research, interviews, and discussions with relevant stakeholders. I would like to express my deep gratitude to everyone who contributed to this paper.

March 2025

HGPI Intractable & Rare Diseases Project

Executive Summary

This discussion paper is structured around five perspectives: social environment, healthcare, family & caregiver support, social participation, and innovation. Each of these perspectives is crucial in the context of policy for intractable and rare diseases, and were major themes raised at the symposium and in interviews conducted in FY2024. Common and central to all five perspectives are the viewpoints of diverse patients and people with lived experience of these diseases. Patients and people living with intractable and rare diseases come from a wide range of backgrounds and lead their daily lives in diverse living environments. We aim to consider the future direction of relevant policies with this reality in mind.

Perspective 1 Fostering a social environment that enables coexistence

From understanding to concrete systems: The need to reflect in policies the *real opinions* of patients and people affected by intractable and rare diseases

The need for activities to raise awareness of intractable and rare diseases among the next generation

The need to understand patients and people with lived experience of intractable and rare diseases who come from diverse backgrounds

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The need to maintain a public health insurance system that supports society while re-evaluating cost-sharing arrangements

The need to establish systems that promote the development and implementation of new diagnostic and treatment technologies sought by patients and people with lived experience

The need for patients and people with lived experience to participate as *planning partners* in the advancement of medical science, healthcare, and policy

Perspective 1

Fostering a Social Environment that Enables Coexistence

From understanding to concrete systems: The need to reflect in policies the real opinions of patients and people affected by intractable and rare diseases

Current policies on intractable and rare diseases are centered around two main themes: medical efforts such as healthcare services, diagnostics, and the promotion of innovation including drug development; and various forms of support grounded in the experiences of people living with rare or intractable diseases in the process from diagnosis to recuperation. In the latter area particularly, policies are such that systems relating to education, welfare, employment, etc., are managed separately, meaning that individuals must overcome through their own efforts any specific issues that fall between those systems. Although there are support and advocacy organizations in various regions seeking to resolve these low-visibility issues, many have insufficient resources and must operate as best they can under the circumstances. Discussions addressing these issues tend to stop at calls for "greater public understanding." However, if we reflect on the history of disability policy—often referenced to in the context of intractable and rare diseases policy—then it is clear that activities are proceeding with the aim of establishing *concrete policies and systems* along with *rights*. One concept that has been gaining traction in recent years is *reasonable accommodation*, as defined in the Convention on the Rights of Persons with Disabilities based on the social model of disability. In the field of intractable and rare diseases, there is also a growing call to move beyond societal understanding and empathy, toward concrete systems based on reasonable accommodation. To achieve this, efforts on the part of stakeholders involved in intractable and rare diseases are crucial. In this regard, the advocacy organizations in various regions are valuable spaces where the real opinions of patients and people with lived experience can be heard. Moreover, in recent years, many such people have been sharing their experiences through blogs and social media. It is hoped that government officials and politicians will not only provide more opportunities for real opinions to be heard, but also take the time to meet with the individuals expressing them and listen to their experiences.

The need for activities to raise awareness of intractable and rare diseases among the next generation

To many people, intractable and rare diseases might, by their very name, seem unfamiliar and irrelevant to them. However, it is estimated that over one million people in Japan live with these diseases, meaning that they could happen to anyone. It is easy to image the challenges that patients and people with lived experience of these diseases face on the path to diagnosis, and the anxiety of coping with worsening symptoms. Furthermore, many of these disease are genetic, and nearly half are diagnosed in infancy or early childhood [i]. Given these realities, it is essential for society as a whole to show interest and understanding toward these people. Government agencies and relevant companies can play in instrumental role in disseminating the information that they provide, but it is important to first increase the total amount of information. In doing so, it is of course necessary to not only ensure the information's accuracy, but also reconsider the appropriateness of terminology such as "intractable disease."

What is particularly needed is outreach to younger generations through education. Education is already being provided on variety of diseases and disabilities, and it is widely reported that these efforts influence not only the students themselves, but their families and others around them. On the other hand, the current national curriculum based on the National Curriculum Standards is already quite dense, making implementation challenging. In recent years, young people have been accessing information via a variety of media such as social media advertising and streaming television. Appealing to younger generations in collaboration with patients and advocate groups is essential for fostering the social environment in the medium to long term.

The need to understand patients and people with lived experience of intractable and rare diseases who come from diverse backgrounds

It goes without saying that “intractable and rare diseases” neither refers to a specific disease nor to a single syndrome. An individual disease is present in each case, and in some instances, a clear disease name or classification has yet to be established. All affected individuals live their daily lives while managing their symptoms, and there are categories besides “patients or people with lived experiences.” There are age-based categories such as children and the elderly, and there are people of other nationalities living in different countries. Some cases may have comorbid chronic disease or disabilities, while others may live alone or in areas with limited social resources such as remote islands or so-called depopulated regions. When considering the needs of people living with intractable and rare diseases, it is also essential to consider the diversity of their circumstances.

Perspective 2

Creating a Healthcare System that Alleviates the Worries of Patients and People with Lived Experience

The issues surrounding healthcare in the context of intractable and rare diseases policy are, as one might expect, diverse. In this discussion, we attempt to organize these challenges from the perspective of the timeline along which citizens receive healthcare services. In Japan, where the public health insurance system is well-established, the process for receiving healthcare services generally follows a common flow. In the majority of cases, this process begins when symptoms come to light. Whether it is an intractable disease or not, we usually respond individually to perceived abnormalities—such as coughing, stomachache, headache, or any deviation from our normal everyday condition—by resting, buying over-the-counter medication, and so forth. If the symptoms worsen or persist, then we consult a physician at a nearby healthcare facility. After various major or minor tests are conducted according to the symptoms, the physician makes a diagnosis and the disease is treated. Occasionally, for strong symptoms that manifest before the disease has been definitively identified, treatment may proceed concurrently with diagnosis. With this context in mind, we will explore the characteristics and challenges associated with medical consultation in the field of intractable and rare diseases.

The need for more diverse access points such as pharmacies, local communities, schools, and workplaces, in order to shorten diagnosis lag

Information on most intractable and rare diseases is rarely encountered, making it difficult for the general public to recognize them at an early stage based solely on their general medical knowledge. If the symptoms worsen or persist, they might be noticed, but many intractable and rare diseases exhibit symptoms that change over time. Determining whether symptoms require medical attention in the first place is in fact a quandary that many people face. In order to detect intractable and rare diseases at an early stage, it is therefore critical to establish systems to provide information and consultation when symptoms are first noticed. In the current healthcare system, there are calls for pharmacists working in pharmacies to take on roles in providing information and consultation. Unlike hospitals, pharmacies do not have long waiting times, making it easy for people who suspect they might be ill to casually consult a pharmacist about whether specialized treatment is necessary. On the other hand, pharmacists also have limited time to spend on such consultation support, and their roles are restricted in the sense that, under the Medical Practitioners' Act, only licensed medical practitioners are permitted to provide diagnoses.

Other opportunities for everyday consultation with healthcare professionals include maternal and child health programs provided by public health nurses during infancy and early childhood, school health programs at educational institutions, and access to occupational physicians at some workplaces. On this note, the Ministry of Education, Culture, Sports, Science and Technology (MEXT) has held review meetings since 2017 on the provision of medical care in schools, promoting collaboration between healthcare professionals and educational institutions, as part of an effort to enhance medical care in educational settings. Thanks to these efforts, the term "school nurse" has become more common, and a growing number of diverse healthcare professionals are exiting medical institutions and becoming active in local communities. We hope that this trend will lead to more points of contact where the public can engage with healthcare so that intractable and rare diseases are detected at an early stage.

In recent years, there has been a demand for improved health literacy and greater individual awareness and knowledge, but—as with intractable and rare diseases—there are still diseases that are difficult to recognize. Therefore, how to increase points of contact with healthcare in daily life remains a crucial perspective for achieving early detection and early intervention.

The need to promote technological innovation and cooperation among healthcare institutions to enable early diagnosis and treatment

Even if a patient manages to visit a healthcare institution at an early stage, further hurdles remain. Intractable and rare diseases are conditions that even medical professionals have limited exposure to. Often their underlying mechanisms

are unknown, and methods of treatment have yet to be established. This can not only delay diagnosis, but even lead to misdiagnosis [ii], which can be a significant source of anxiety for patients and affected individuals.

The Initiative on Rare and Undiagnosed Diseases (IRUD), led by the Japan Agency for Medical Research and Development (AMED), is an initiative to reduce the number of undiagnosed diseases by analyzing patients' genetic information to search for diagnostic clues. The initiative promotes early diagnosis of rare diseases, identifies causative genes underlying undiagnosed diseases, and establishes new disease concepts. For cases where a condition is identified and diagnosed as a known disease, there are specialists for the various respective diseases. For such cases, a system is in place to ensure that patients can receive early consultation with professionals specializing in the various diseases, in accordance with the Basic Policy for Comprehensively Promoting Medical Care for Patients with Intractable Diseases. Looking ahead, there are growing expectations for the realization of earlier diagnosis and treatment through several means: the wider adoption of scientifically proven newborn mass screening; advancements in diagnostic prediction and diagnostic technologies including the use of artificial intelligence (AI), that help primary care physicians to detect potential intractable and rare diseases at the stage of the initial consultation; smoother referrals to specialists thereafter; and the provision of information and educational opportunities for general physicians to support this process. To achieve this, it is essential to promote research and development, make continuous investments in nurturing researchers, and strengthen systems for public–private collaboration.

The need to design systems based on a life-course approach that accommodates individual characteristics and long-term medical care

In considering the lives of people living with intractable and rare diseases from a life-course perspective, one essential element in policy is transitional care. If a disease appears in infancy or childhood, the first point of consultation is often a pediatrics department. Long-term treatment from childhood involves not only changes in symptoms but also simultaneous growth and development, requiring ongoing adjustments in treatment methods and care to match the patient's physical and mental development. Additionally, in long-term care starting in childhood, it is important to promote the independence of the individuals living with intractable and rare diseases, while also providing support for those in caregiving roles. Long-term treatment brings a variety of challenges that must be resolved, such as transferal from pediatrics to a number of adult departments, and switching to a different medical expense subsidy program (e.g., from a specified chronic pediatric disease program to a designated intractable disease program).

Some progress has been made in this area: a survey conducted in 2023 revealed that the ease of transition can vary depending on the disease group, and attempted to categorize the patterns of transition. However, factors such as the age at which transition support begins and ends, and the duration required to complete the transition, can differ significantly even within the same disease group [iii], highlighting the limitations of addressing these challenges solely within the framework of "intractable and rare diseases." Approximately 800 diseases under 16 disease groups are recognized as specified chronic pediatric diseases, and over 300 diseases are recognized as designated intractable diseases—and the latter figure continues to grow. Both categories contain conditions with individual, disease-specific characteristics. Many cases involve symptoms or disease affecting multiple organs, and the fact that nominal severity of the disease does not always match the actual level of support required in daily life presents an additional challenge.

From the perspective or people with living with intractable or rare diseases and their families, issues relating to medical and care expenses—which greatly impact daily life and continued access to medical consultation—are urgent issues in transitional care. Measures for specified chronic pediatric diseases and designated intractable diseases are based on different laws with different objectives, and as such, their continuity is not necessarily adequate. However, what is important for people living with intractable and rare diseases is the ability to seamlessly receive ongoing support regardless of the system they fall under. Currently, subsidies for specified chronic pediatric diseases are limited by patient age, and it has been pointed out that treatment is sometimes discontinued once that age limit is reached, regardless of whether continued treatment or monitoring is needed. To ensure that people with intractable or rare diseases can continue receiving necessary treatment and live with peace of mind, we face the endless challenge of how to strike a balance between ensuring uniformity and fairness within the system, and providing personalization and flexibility to meet individual needs.

Perspective 3

Developing Policies that Uphold the Dignity of Families and Caregivers

The need to shed light on the circumstances of families and caregivers, help to reduce their burdens, and support social participation and self-actualization

In the care of individuals living with intractable and rare diseases, it is typically the parents or guardians who take on the primary caregiving role, particularly when the disease appears in childhood or earlier. For parents and guardians, raising a child living with an intractable or rare disease means carrying the weight of a great many problems, concerns, and anxieties. In addition to the normal responsibilities of parenting, these include maintaining the child's life, planning for education and employment, and coping with the medical and financial burden of long-term treatment. With physical and mental growth and development, and effective symptom management, it is possible for such children to lead independent lives. As a result, parents and guardians face a wide range of challenges over many years, corresponding to the stage of their child's development.

The dilemma between the responsibility of providing care and encouraging independence is not unique to intractable and rare diseases. For example, the cerebral palsy rights movement of the 1970s, which pioneered the disability rights movement in Japan, was sparked by the struggles of young cerebral palsy patients and the mothers who cared for them. In Japan, where social policy tends to family-oriented, the role of the family and informal care tends to be emphasized in all aspects from child-rearing to elder care, with much of the burden falling on women. The vision of the independent living movement for people with disabilities is for them to live independently in their communities, not in institutions, based on their own decisions, with support such as care assistance, regardless of the severity of their disabilities. As a result, families and caregivers can also live their own lives. In recent years, local governments have begun to enact "caregiver ordinances," reflecting a renewed effort to frame the anxieties and challenges faced by families and caregivers not merely as family matters, but as societal issues.

Intractable and rare diseases, due to their treatment-resistant nature, require a patient to live with them long-term, and are characterized by a tendency to present a different condition or symptoms depending on growth or environment, regardless of the patient's intentions. This raises concerns about such fluctuations significantly impacting the patient's independence. For example, even if an individual learns to effectively control their symptoms and engage in social activities such as school or work, a change in their condition could force them to reassess how they deal with their symptoms. If this cycle occurs repeatedly, it can decrease the individual's motivation to cope with their symptoms, and potentially reduce their motivation to participate in social activities. To assist such individuals, various support programs have been established, such as Intractable Disease Consultation Support Centers under the Intractable Diseases Act, independent support initiatives for children with specified chronic pediatric diseases, and independent support under the Act for the Comprehensive Support of Persons with Disabilities.

On the other hand, support for informal care providers such as family and caregivers is rarely mentioned explicitly. We therefore hope that efforts will be made to clarify the situation faced by family and caregivers of people living with intractable and rare diseases, acknowledge their existence, and construct support systems as required. There is a need for systems that enable people living with intractable and rare diseases to live as independently as possible without relying on informal care from family and caregivers. However, given the strong tendency in Japan for caregiving to be family-based, the reality is that the role of families and caregivers inevitably becomes significant—especially in the case of rare and intractable diseases, where even a minor mistake can have life-threatening consequences, and where points requiring attention change over time. As humans beings grow, they take on various roles and strive for self-actualization. When family members and caregivers—whose lives tend to be relatively occupied by child-rearing and caregiving roles—are able

to participate in society and pursue self-actualization, it also contributes to the child's growth. To help children to safely achieve independence, it is necessary to establish a care system that includes the family, and to build an environment together with society as a whole that accommodates intractable and rare diseases.

Perspective 4

Ensuring and Enhancing Opportunities for Social Participation (Education and Employment)

The need to guarantee diverse learning environments for children living with intractable and rare diseases, and to build flexible education systems

Rights and systems relating to school education have been implemented under various laws and measures. The Convention on the Rights of the Child, adopted by the United Nations in 1989, affirms the rights of all children to receive an education, and calls for mandatory access to elementary education, as well as for opportunities to receive secondary and higher education. The provision of educational opportunities is also addressed in various treaties, laws, and regulations such as the Convention on the Rights of Persons with Disabilities, the Basic Act on Education, the Child Welfare Act, and the Basic Act on Children's Policy. To protect these rights and ensure access to opportunities, the Act for Eliminating Discrimination against Persons with Disabilities requires that necessary and reasonable accommodation be provided to remove social barriers in accordance with the gender, age, and disability characteristics of school-aged children with disabilities.

However, according to a survey by MEXT, when prefectural and municipal boards of education were asked to what extend they provided initiatives or support for schools attended by children receiving medical treatment, 82% reported providing support to special needs schools, while support for elementary, junior high, and senior high schools all fell short of 50%[iv]. The survey results also indicated that at present, systems for accepting children and students living with intractable and rare diseases are not fully established in all regions, and that individual regions or schools may lack sufficient experience or knowledge if a need suddenly arises.

It would be ideal if children and students with diseases that potentially change over time can choose, based on their circumstances, from multiple school and system options—for example, in terms of school and class types, the main options would be special needs schools, special needs classes, and regular classes. In terms of systems, options might include tsūkyū classes (resource rooms) and on-demand lessons for children and students unable to attend school due to home medical care. Today, an increasing number of schools are offering inclusive education, and it is expected that systems will improve as ICT is adopted. On the other hand, teacher shortages and excessive teacher workloads are becoming apparent even in regular classrooms, so it should be noted that under the current education system, the risk of schools' burdens increasing cannot be ruled out.

The need to establish diverse employment options to support the economic independence of patients and people with lived experience

Employment is one of the main avenues for social participation after completing education. It is important not only for establishing economic independence, but also for increasing self-esteem. School curricula include programs to prepare students for employment, and some students begin exploring work styles and workplaces at an early stage to find employment after graduation. Existing support systems include disability welfare services under the Act for the Comprehensive Support of Persons with Disabilities, and employment support services provided as part of self-reliance support services for children with specified chronic pediatric diseases. Furthermore, under the disabled worker hiring provisions of the Act to Facilitate the Employment of Persons with Disabilities, companies are required to hire a minimum mandatory proportion of individuals with physical, intellectual, or mental disabilities (known as the "statutory employment rate"). At present, individuals living with intractable and rare diseases are not included in this framework, but there is potential to consider including them in the future.

On the other hand, not all individuals living with intractable and rare diseases hold a disability passbook, and many cases do not fall under the traditional framework of disability employment. For example, thanks to advances in medical technology, the symptoms of many diseases can be stabilized without resulting in physical disability. Yet in some cases, symptoms can still cause limitations in daily life if the condition does not qualify as a disability. Similarly, individuals

with conditions involving susceptibility to infection may require daily infection control measures even if they are not recognized as having a disability. People may also face other diverse hardships, such as pain that is difficult to explain objectively, or chronic fatigue. Initiatives to promote employment among people living with intractable and rare diseases under the current system include the establishment of Intractable Disease Consultation Support Centers and assignment of Intractable Disease Employment Support Coordinators by prefectural governments under the Intractable Diseases Act. However, attention is being drawn to a shortage of facilities and personnel. In 2018, a *Management Manual for the Employment of People with Intractable Diseases* was published for employers, to promote understanding of intractable diseases and highlight points of concern related to employment.

Since April 1, 2024, businesses have been required by law to provide reasonable accommodation for people with disabilities. Looking ahead, it is hoped that employment options for patients and people with lived experience will be increased by expanding employment support systems for people with intractable and rare diseases, while considering the use of existing disability employment frameworks.

The need for employment environments that enable medium- to long-term career-building while balancing treatment and work

Once employment is secured and work begins, new challenges arise such as maintaining employment in line with personal changes, and building a career through promotion or job changes. The most notable system for supporting continued employment is the workplace adaptation assistant (job coach) support program. When issues in workplace adaptation arise, job coaches provide specialized support tailored to an individual's disability characteristics, with the aim of helping the disabled individual adapt to their work environment. Furthermore, a medical fee system, known as the Medical Treatment and Work Balance Support Guidance Fee, was introduced in 2018, enabling doctors to be recognized for actively supporting the employment of people living with intractable or rare diseases. In addition, changes in work styles brought about by the COVID-19 pandemic have encouraged companies to develop more flexible working environments—such as remote work and four-day workweeks—and many have begun to independently implement their own initiatives to support continued employment.

Yet in today's society, where various forms of career-building such as job-switching or starting a business are being explored, it is important to support not only continued employment, but also career building. An example of this is the Career Development Promotion Grant (Permanent Employment for Persons with Disabilities Course), launched in 2021 to promote the employment and workplace retention of individuals with disabilities, which subsidizes employers who convert fixed-term employees with disabilities into permanent employees. Since support for job changes is limited, each consultation must be handled carefully and individually. However, this can lead to a problem where if a large number of people are seeking help, changes in symptoms or circumstances can occur while waiting for one's turn, causing people to leave their jobs without waiting.

Intractable and rare diseases are characterized by symptoms and conditions that change over time, as well as a need for ongoing outpatient visits, which all pose a significant barrier to continued employment and career-building. The main challenge for an individual living with these diseases is to balance work and career-building with treatment. To achieve this, it is important for the primary care physician and workplace to collaborate closely and make ongoing adjustments that accommodate the individual. At present, employment and career-building for people living with intractable and rare diseases is often discussed within the framework of disability employment. However, it is hoped that future discussions and systems will take into account the challenges specific to intractable and rare diseases.

Perspective 5

Realizing a Policy Environment where Patients and People with Lived Experience Can Benefit from Innovation

The need to maintain a public health insurance system that supports society while re-evaluating cost-sharing arrangements

Intractable and rare diseases can affect anyone, and once diagnosed, long-term treatment is often required, entailing a significant financial burden. Sharing this lifestyle risk across society as a whole is the basic idea of the public health insurance system, and intractable and rare diseases are precisely the types of conditions that should be supported within this framework. When patients and people with lived experience of intractable and rare diseases are supported by such systems, it fosters a sense of security and solidarity across society as a whole.

On the other hand, it is widely known that the public health insurance system is under financial strain. However, it is unacceptable to attempt to financially sustain it by shifting the burden of expensive treatments or medications to private medical insurance, or increasing out-of-pocket costs for patients, as it could exacerbate healthcare inequalities caused by economic factors. At the same time, there are limits to what can be provided by the patchwork financial resources of the existing system—which is why bold systemic reform is necessary. The present paper is not intended to directly address reform of the public health insurance system, so discussion will be kept to a minimum. However, one idea could be to increase out-of-pocket costs for certain drugs that generally require a prescription to buy despite not being classified as prescription drugs (so-called OTC-like drugs). Such a policy could be combined with burden reduction measures such as expanding the self-medication tax system, or introducing a fixed deductible as used in auto insurance, in order to help gain public understanding.

The purpose of Japan's public health insurance system, including the high-cost medical expense system, as stated in Article 1 of the Health Insurance Act, is "contributing to the stability of lives and the improvement of welfare of the people." Given this fact, broad public discussion and bold political decisions are necessary to ensure that it can sustainably provide support in times of need.

The need to establish systems that promote the development and implementation of new diagnostic and treatment technologies sought by patients and people with lived experience

As noted above, if the public health insurance can continue to support patients and people with lived experience of intractable and rare diseases, it opens the door to them benefiting from new diagnostic and treatment technologies in a stable manner. Several considerations for achieving this will now be described.

Firstly, the development of diagnostic and treatment technologies requires the construction of systems to collect the patient data upon which they will be based. In the case of intractable and rare diseases, research progress is hindered by the small number of patients overall, which limits the amount of data that individual research institutions hold. Public-private collaboration is therefore necessary to develop a database convenient for research use, that manages and provides not only medical information such as electronic patient records, but also personal health records (PHRs) and genomic data. In a public opinion survey conducted by HGPI, the most commonly supported purpose for the use of anonymized medical data was "the development of new treatments or therapeutics." [v] In planning such systems, the participation of patients and people with lived experience is essential. By learning about the pros and cons of these initiatives, understanding them, and offering constructive criticism, surely they can build an equal and collaborative relationship with researchers.

Policy support is also essential to promote public-private development, including connecting basic research conducted at universities and research institutions with product development in the private sector. Moreover, the existing drug pricing system has difficulties sustaining research and ensuring stable supplies in the field of intractable and rare diseases, leading to problems of drug lag and drug loss. Improving the clinical trial process based on scientific evidence, including better access for patients and people with lived experience, along with the use of conditional approval systems, is

expected to enhance access to innovative treatments.

Another important consideration is the perceived therapeutic effectiveness of a treatment among patients, people with lived experience, doctors, and families after a drug is brought to market. Surely taking into account such contextual evidence and secondary outcomes in drug and medical supply pricing is more considerate from the perspective of patients and people with lived experience.

The need for patients and people with lived experience to participate as planning partners in the advancement of medical science, healthcare, and policy

Today, patient and public involvement (PPI) is seen across a wide range of diseases, and the field of intractable and rare diseases has played a pioneering role in bringing this about. Going forward, it is expected that engagement will move beyond simply *making patients' voices heard* to a model of *co-creation*. The systemic reforms described above can only be realized when patients and people with lived experience are recognized not only as the recipients of medical services, but as proactive participants in sustaining and constructing these systems.

As noted earlier, PPI is essential for the development of new diagnostic and treatment technologies. One challenge in addressing intractable and rare diseases is that patients and people with lived experience are not always in close contact with healthcare, precisely because treatments have not been established. We hold hopes for initiatives, exemplified by Osaka University's Commons Project[vi], that draw on the difficulties faced by patients and people with lived experience, develop them into objective evidence, and apply them in R&D and policymaking.

However, the operating environment for patient and advocacy groups is far from sufficient. Many of these groups are run by a small number of people and have extremely limited financial resources. Most focus primarily on peer support, and only a few are involved in research and development or policy advocacy. HGPI is well aware that one of our missions is to work to help raise the value of these civil society organizations. At the same time, we anticipate that government, industry, and researchers will also provide impartial multifaceted support for the activities of patient and advocacy groups, not only financially, but also through human and intellectual resources.

i) Global Genes website: https://globalgenes.org/rare-disease-facts/ (last accessed on March 4, 2025)
ii) Japan Pharmaceutical Manufacturers Association Intractable and Rare Diseases Task Force (2023), *Survey of Anxieties faced by Patients with Intractable and Rare Diseases*
iii) PwC Consulting LLC (2023), *Survey Report on the State of Transitional Care Support Systems under the FY2022 Program for Promoting Systems for Intractable Diseases*
iv) Special Needs Education Division, Elementary and Secondary Education Bureau, MEXT (2023), *FY2022 Survey Results on the Actual Conditions of Children Receiving Medical Treatment*
v) HGPI (2025), *The 2025 Public Opinion Survey on Healthcare in Japan*
vi) Osaka University Graduate School of Medicine, Biomedical Ethics and Public Policy, *Building a Commons for Evidence Generation Using ICT in Medical Science and Healthcare and Applying It to Policy* https://www.med.osaka-u.ac.jp/pub/eth/research-project/ (last accessed on March 4, 2025)

Health and Global Policy Institute: Guidelines on Grants and Contributions

As an independent, non-profit, non-partisan private think tank, HGPI complies with the following guidelines relating to the receipt of grants and contributions.

1. Approval of Mission

The mission of HGPI is to improve the civic mind and individuals' well-being, and to foster a sustainable healthy community by shaping ideas and values, reaching out to global needs, and catalyzing society for impact. The activities of the Institute are supported by organizations and individuals who are in agreement with this mission.

2. Political Neutrality

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