5. Conclusion

In this survey, we identified challenges from the perspective of healthcare professionals in five areas (1. Basic and applied research, 2. Development and clinical trials, 3. Diagnosis, 4. Treatment and prognosis management, 5. Disease awareness) and analyzed their underlying causes. Furthermore, we clarified expectations of each stakeholder (pharmaceutical industry, academic societies, patient advocacy groups, and the government) and organized the ideal form for improving medical care for rare diseases, specific response measures and their roles.

As detailed in this report, there are numerous challenges in the diagnosis and treatment of rare diseases. Notably, delayed diagnoses, limited treatment options, and difficulties for patients in accessing information are among the key challenges. Overcoming these challenges necessitates the cooperation and collaboration of all stakeholders, not only in enhancing diagnostic techniques and developing new treatments but also in strengthening patient support. Additionally, it is crucial to deepen public understanding of the challenges associated with rare diseases through awareness campaigns.

This survey also targeted healthcare professionals who have been leading the way in rare disease medicine in specific medical departments and disease areas in Japan. By expanding and delving deeper into the survey population in the future, we aim to further concretize the challenges and expectations identified in this survey and advance towards realizing the envisioned ideal state.

We, IRUD, RDCJ and JPMA, express our respect for the energetic activities and significant contributions of all those involved to date. We are committed to working in collaboration with stakeholders to address the diverse challenges surrounding healthcare professionals identified in this survey, with the goal of creating a more livable society for patients with rare diseases and their families.

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Rare Disease Consortium Japan

Rare Disease Consortium Japan was established on February 29, 2024, as an organization that promotes medical research and drug discovery for rare diseases, with collaboration amongst industry, academia, the public, and private sectors at its core with the aim of realizing patient-centered medical services. The fact that we were able to conduct the 'Survey on the challenges faced by healthcare professionals in rare diseases' in collaboration with the JPMA and IRUD was a valuable opportunity to give momentum to our organization's activities. We would like to express our sincere gratitude to all those involved who provided their support and cooperation in carrying out this project.

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JPMA

Rare and Intractable Disease Task Force

Japan Pharmaceutical Manufacturers Association (JPMA) organized Rare and Intractable Disease Task Force in 2021 to pay tribute to the dedicated activities and significant contributions of those involved in intractable and rare diseases and to play a part in this effort. In 2023, we executed and published a ‘Survey on the challenges faced by healthcare professionals in rare diseases’, compiled a recommendation on rare and intractable diseases and are working with relevant parties to resolve these challenges. We would like to express our sincere gratitude to all those involved for their support and cooperation in conducting the survey on the challenges facing healthcare professionals in rare diseases and in compiling this report.

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EY Parthenon

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EY Parthenon Health & Life Sciences team has developed feasible strategies that go beyond the traditional framework of consulting to ensure sustainable development of healthcare system for the next 100 years and provides optimal services to all stakeholders across industries. Leveraging our global network and scale, members with diverse expertise we have organically collaborated with a wide range of stakeholders to conceive and implement problem-solving solutions that transcend the boundaries of private, public and academic sectors. In this survey, we supported the planning and management of the survey together with IRUD, RDCJ and JPMA. We would like to express our sincere gratitude to all involved parties, and we will continue to aim to build a better working world for all those involved in rare diseases.

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