News Release

February 25, 2026
CareNet, Inc.
Macromill Carenet, Inc.

[In Recognition of Rare Disease Day (RDD), Survey Conducted Among General Medicine Physicians]
Approximately 80% of General Medicine Physicians Report Keeping "Rare Diseases" in Mind as Potential Diagnoses in Daily Practice
— Strong Motivation for Early Detection and the Need for Diagnostic Support Systems Revealed —

CareNet, Inc. (Head Office: Chiyoda-ku, Tokyo; President and CEO: Katsuhiro Fujii) and Macromill Carenet, Inc. (Head Office: Minato-ku, Tokyo; President and CEO: Shigeru Tokuda) conducted a survey to assess awareness of rare diseases among 108 general medicine physicians in conjunction with Rare Disease Day (RDD) on February 28.

At the outset of the survey, respondents were asked about the time required to reach a confirmed diagnosis of a rare disease (defined in Japan as a disease affecting fewer than 50,000 patients). Ninety percent of general medicine physicians responded that rare diseases are more prone to diagnostic delays than common diseases. Against this backdrop, the survey examined how general medicine physicians approach rare diseases in their daily clinical practice.

More than 80% of general medicine physicians reported that they are "aware of rare diseases as potential diagnoses" in daily practice. Approximately 70% viewed themselves as "initial evaluators and gatekeepers", serving as "key contributors to the diagnostic process" for rare diseases. These findings suggest that many general medicine physicians keep rare diseases in mind during routine clinical practice.

On the other hand, approximately one in four general medicine physicians reported

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having experience diagnosing at least one rare disease per year. Additionally, 76% of general medicine physicians responded that “they can contribute to the early detection of rare diseases,” suggesting that even in the field of rare diseases, where opportunities for clinical experience are limited, general medicine physicians have a strong recognition of their role and a clear intention to contribute to early diagnosis.

Comment from Professor Tetsuhiro Maeno, Chair, Department of General Medicine and Primary Care, University of Tsukuba Hospital

Commenting on the survey results, Professor Tetsuhiro Maeno, Chair of the Department of General Medicine and Primary Care at the University of Tsukuba Hospital, stated:

“Although the number of patients affected by each rare disease is limited, the total number of rare disease types exceeds several thousand. Consequently, the overall patient population is by no means ‘rare’ in scale. However, owing to the high level of specialization required and the diversity of symptoms, it is not uncommon for patients to face social challenges, as it can take several years on average to reach an appropriate diagnosis.

Against this backdrop, the findings of this survey suggest that many general medicine physicians are mindful of rare diseases in their daily clinical practice and position themselves as ‘gatekeepers’ in the diagnostic process, demonstrating a strong commitment to contributing to earlier detection.

However, because opportunities to encounter rare diseases in routine practice are limited, general medicine physicians may find it difficult to accumulate hands-on experience. Consequently, even when physicians are aware of the existence of rare diseases, it can be challenging to suspect them while managing a large volume of common disease cases in daily practice, which may contribute to diagnostic delays.

In addition to enhancing the expertise of individual physicians, it is important to advance the development of systems that enable the aggregation and sharing of knowledge.”

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Key Survey Findings

1. More than 80% of General Medicine Physicians Are Aware of Rare Diseases as Potential Diagnoses in Daily Practice

When asked about the “frequency with which rare diseases are considered as part of differential diagnosis in routine practice”, 14% responded that they “always consider” them, while 68% responded that they “consider them depending on the case or symptoms.” In total, more than 80% of general medicine physicians reported that they consider rare diseases during their daily clinical work.

Although rare diseases are individually uncommon, these results suggest that many general medicine physicians consider them a possibility in routine clinical practice.

2. Many General Medicine Physicians Recognize Their Role as the Starting Point in Rare Disease Diagnosis and Believe They Can Contribute to Early Detection

When asked about “the most important role of general medicine physicians in reducing diagnostic delays for rare diseases,” 46% identified themselves as “initial evaluators and gatekeepers to the diagnostic process,” and 24% selected “management of the diagnostic process.”

In response to the question “Do you believe general medicine physicians can contribute to the early detection of rare diseases?”, 76% responded that they “can contribute.” These findings indicate strong awareness among general medicine physicians of their role as the “starting point” in rare disease diagnosis and as “key contributors” within the overall diagnostic process, reflecting a high level of motivation to support earlier diagnosis.

3. Approximately One in Four General Medicine Physicians Has Experience in the Diagnosis of at Least One Rare Disease Case per Year

In practice, 24% of general medicine physicians reported having been involved in the diagnostic process for at least one rare disease case per year. Although physicians are aware of rare diseases and recognize their role, opportunities to encounter such

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cases in daily clinical settings remain limited, suggesting that rare disease diagnosis is an area in which practical experience is difficult to accumulate.

4. Ninety Percent of General Medicine Physicians Perceive That “Rare Diseases Tend to Take Longer to Diagnose”

When asked whether, compared to common diseases, “rare diseases tend to involve delays from a patient’s initial medical consultation to a confirmed diagnosis,” 90% of general medicine physicians responded that delays are more likely to occur.

5. Early Diagnosis of Rare Diseases Requires Both Enhancing Clinical Expertise Among General Medicine Physicians and the Development of Diagnostic Support Systems

When asked what types of support are necessary to promote the early diagnosis of rare diseases, the most frequently cited responses were “enhancing clinical expertise of general medicine physicians” (63%) and “developing diagnostic support tools and clinical guidelines” (49%).

These findings suggest that in rare disease diagnosis, solutions cannot rely solely on the individual efforts or experience of physicians. General medicine physicians recognize the importance of establishing systems that support knowledge sharing and clinical decision-making, in addition to enhancing individual competencies.

Future Outlook

This survey revealed both the strong commitment of general medicine physicians toward rare diseases and the unique challenges in this field, particularly the difficulty of accumulating sufficient knowledge based solely on individual clinical experience.

CareNet and Macromill CareNet, as platforms for physicians and healthcare professionals, have supported clinical decision-making in real-world practice by aggregating and disseminating knowledge related to rare and intractable diseases. By leveraging the insights gained from this survey, we will further strengthen our role as a “knowledge-integration hub,” connecting individual clinical experiences with

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collective expertise across the healthcare community.

More than 200 rare diseases are currently covered in the CareNet.com “Rare Disease Library,” which provides fundamental information on diagnosis and treatment (available to physician members only):
https://www.carenet.com/report/library/general/rare/index.html

The official CareNet LinkedIn account also distributes content under the theme “Understanding Rare and Intractable Diseases”.
https://www.linkedin.com/company/carenet-inc/posts/

About Macromill Carenet, Inc.

Company Name: Macromill Carenet, Inc.
Representative: Shigeru Tokuda, President and CEO
Location: Shinagawa East One Tower 11F, 2-16-1 Konan, Minato-ku, Tokyo, Japan
Established: December 2014
Capital: JPY 45 million
Business Description: Market research specializing in the healthcare sector
Website: https://www.macromillcarenet.jp/

About CareNet Group

The CareNet Group seeks to "support medical professionals and move the future of medicine through knowledge, passion, and action." It conducts its business on CareNet (https://www.carenet.com/), a platform with over 240,000 physician members. We provide a wide range of specialized services in the medical and pharmaceutical fields, ranging from healthcare human resources, education, and management to supporting new drug development, clinical trials, and information dissemination.
Please refer to https://carenet.co.jp/ for an overview of CareNet.
Recruitment information is available at https://carenet.co.jp/recruit

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Reference Materials: Survey Overview and Results

[Survey Overview]

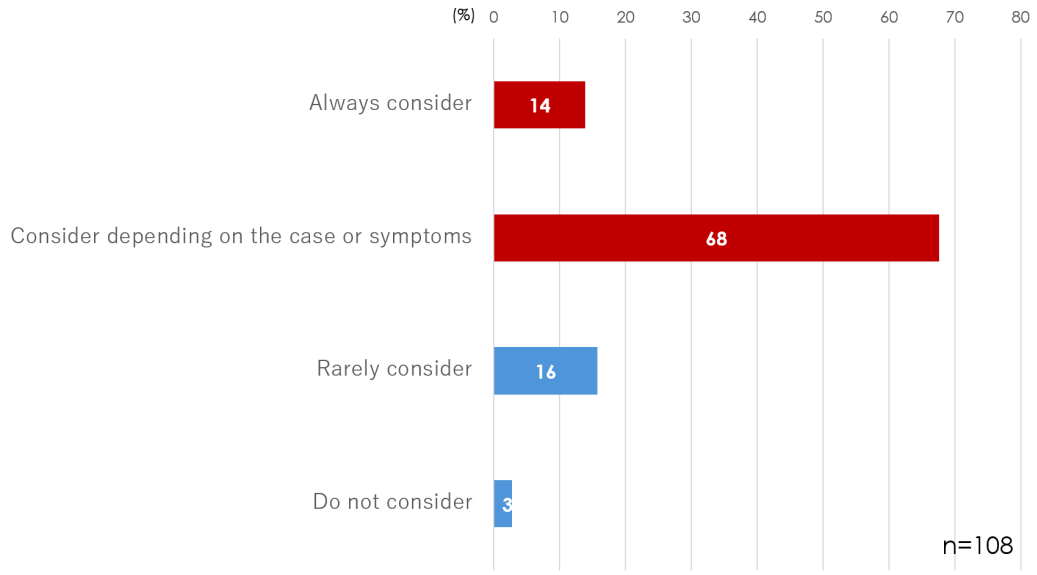
An internet-based survey was conducted among general medicine physicians working in general medicine departments to assess their awareness of rare diseases.

Survey Period: December 26–31, 2025
Survey Method: Online survey
Respondents: 108 physicians working in general medicine departments
Survey Panel: Physicians registered with CareNet.com

[Survey Results and Charts]

<More than 80% of General Medicine Physicians Consider Rare Diseases in Differential Diagnosis During Routine Practice>

Q. How frequently do you consider rare diseases as part of your differential diagnosis in daily clinical practice?



Combining “Always consider” (14%) and “Consider depending on the case or symptoms” (68%), a total of 82% of general medicine physicians reported that they take rare diseases into account in their clinical practice.

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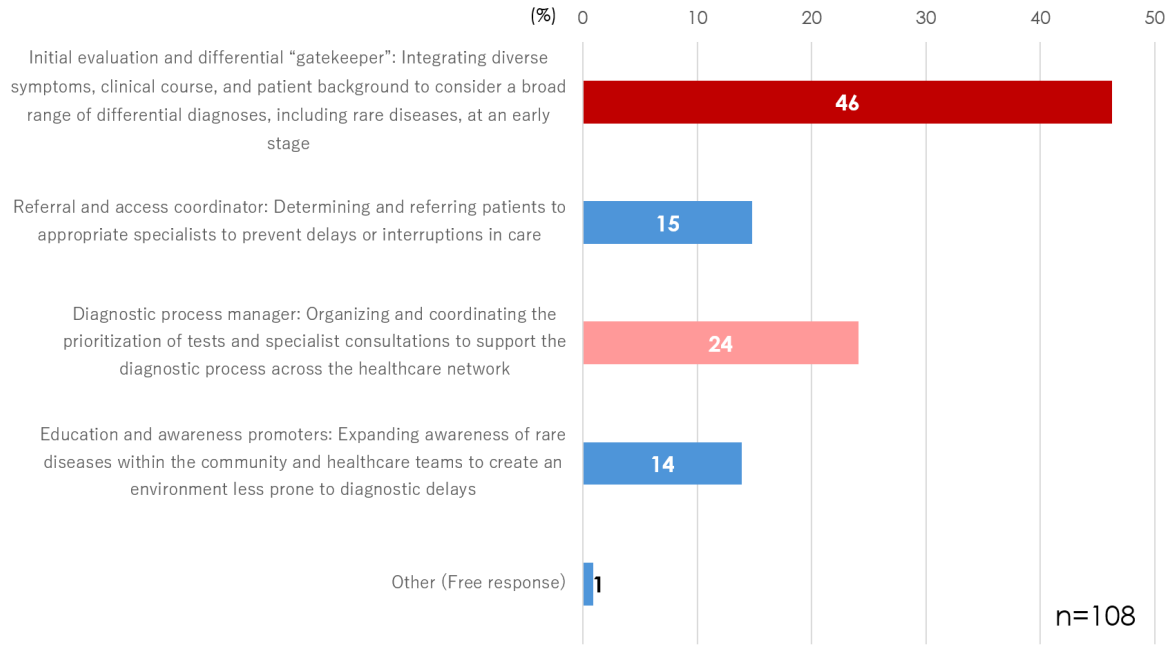
<Many General Medicine Physicians Recognize Their Role as the Starting Point in Rare Disease Diagnosis and Believe They Can Contribute to Early Detection>

Q. To reduce diagnostic delays in rare diseases, which single role do you believe is most important for general medicine physicians?



Forty-six percent selected “initial evaluation and differential gatekeeper,” and 24% selected “diagnostic process manager.”

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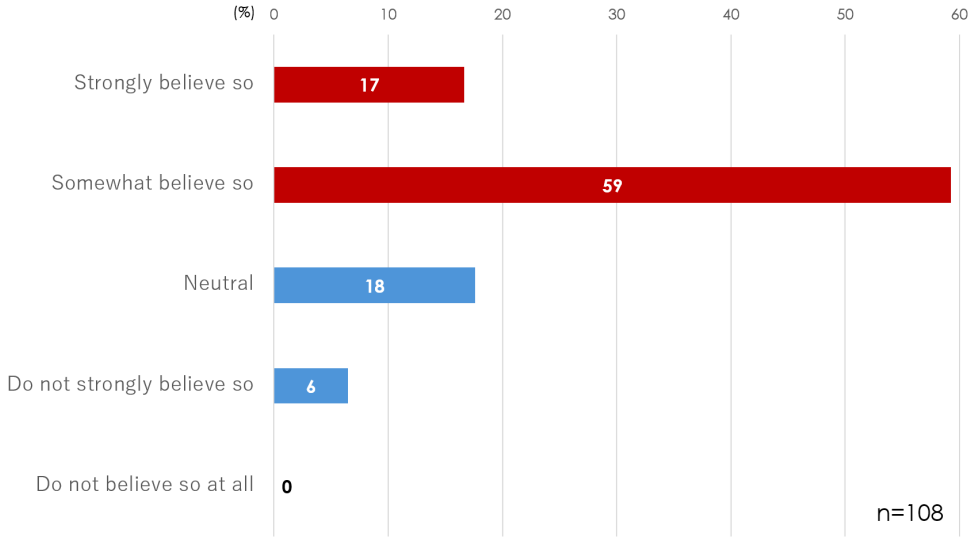
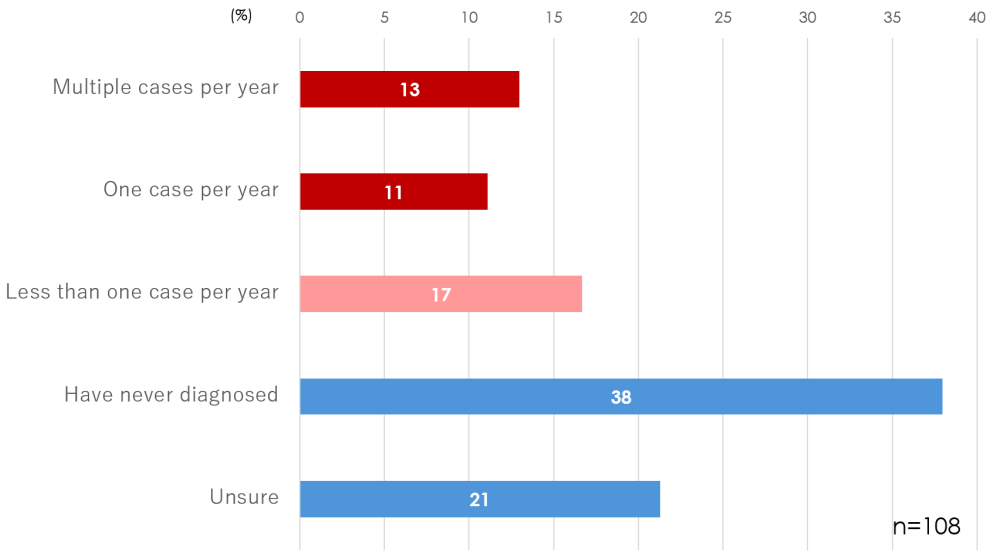
Q. Do you believe general medicine physicians can contribute to the early detection of rare diseases?



In total, 76% (17% + 59%) responded that general medicine physicians could contribute to early detection.

<Approximately One in Four Physicians Has Been Involved in the Diagnosis of at Least One Rare Disease Case per Year>

Q. As a general medicine physician, approximately how many rare disease cases do you diagnose per year (including cases in which you were involved in the diagnostic process)?



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Twenty-four percent (13% + 11%) reported being involved in at least one rare disease diagnosis per year. Meanwhile, more than half responded either “have never diagnosed” (38%) or “unsure” (21%).

Q. For those who selected “multiple cases per year,” “one case per year,” or “less than one case per year,” please specify the rare diseases involved (free response).

Disease Name	Count
Familial Mediterranean Fever (including “Mediterranean fever”)	7
EGPA (Eosinophilic Granulomatosis with Polyangiitis), Fabry disease, HAE (Hereditary Angioedema)	3
Adult-onset Still’s disease, pemphigus, Osler disease (Hereditary Hemorrhagic Telangiectasia), cardiac amyloidosis, VEXAS syndrome, dermatomyositis (including MDA5-positive), anti-NMDA receptor encephalitis, adrenal insufficiency	2
ALS (Amyotrophic Lateral Sclerosis), CJD (Creutzfeldt–Jakob disease), Dravet syndrome, PSP (Progressive Supranuclear Palsy), SLE (Systemic Lupus Erythematosus), ANCA-associated vasculitis, TAFRO syndrome, Kimura disease, bullous pemphigoid, PFAPA syndrome, stiff-person syndrome, idiopathic ureteral rupture, polysplenia, familial hypercholesterolemia, Parkinson’s disease, hypoparathyroidism, splenic lymphangioma, HIT (Heparin-Induced Thrombocytopenia), post-streptococcal reactive arthritis, cancer of unknown primary, tetanus, TTP (Thrombotic Thrombocytopenic Purpura), FGF23-related hypophosphatemia, sarcoidosis, soft tissue sarcoma, isolated ACTH deficiency, JMML (Juvenile Myelomonocytic Leukemia), giant cell arteritis, paroxysmal cold hemoglobinuria, small intestine cancer, Bickerstaff brainstem encephalitis, Japanese encephalitis	1

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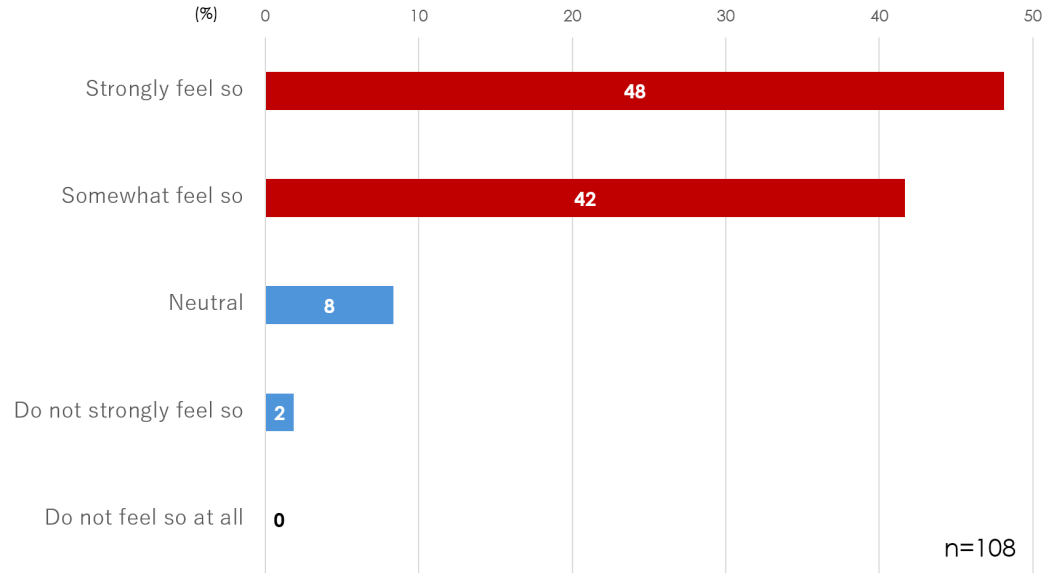
<Ninety Percent of General Medicine Physicians Feel That Rare Diseases Tend to Take Longer to Diagnose>

Q. Compared to other diseases, do you feel that rare diseases (defined in Japan as affecting fewer than 50,000 patients) are more likely to experience delays from initial consultation to confirmed diagnosis?



Overall, 90% (48% + 42%) responded that rare diseases were more likely to involve diagnostic delays.

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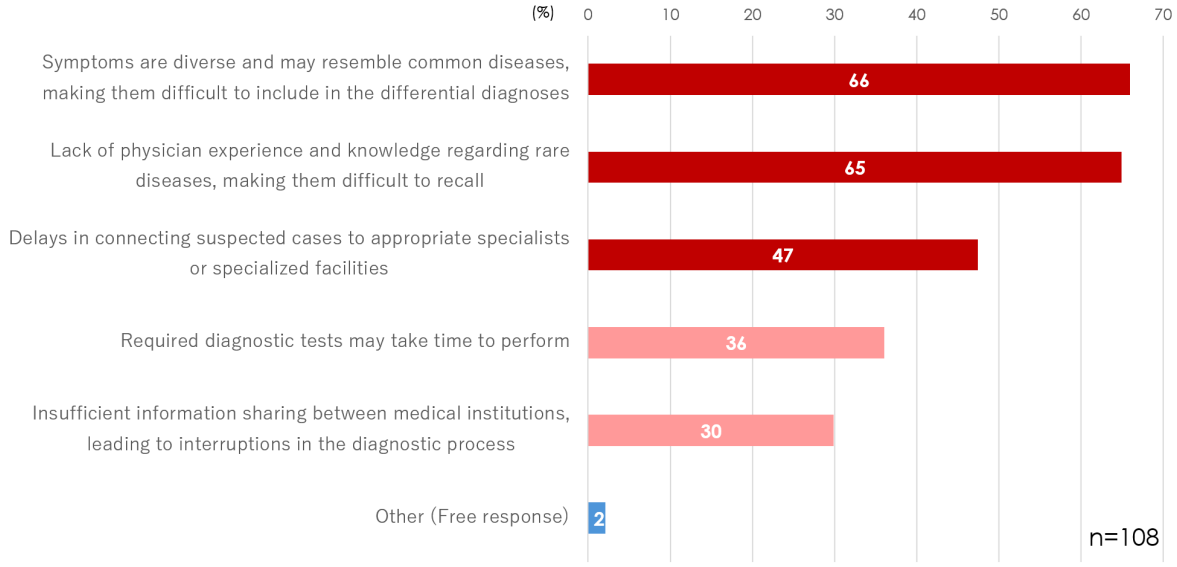
Q. For those who answered “strongly feel so” or “somewhat feel so,” what do you believe are the reasons? (Multiple answers allowed)



The most commonly cited reasons were difficulty in identifying rare diseases as differential diagnoses due to symptom diversity (66%), insufficient physician experience and knowledge (65%), and delays in referral to appropriate specialists or facilities (47%).

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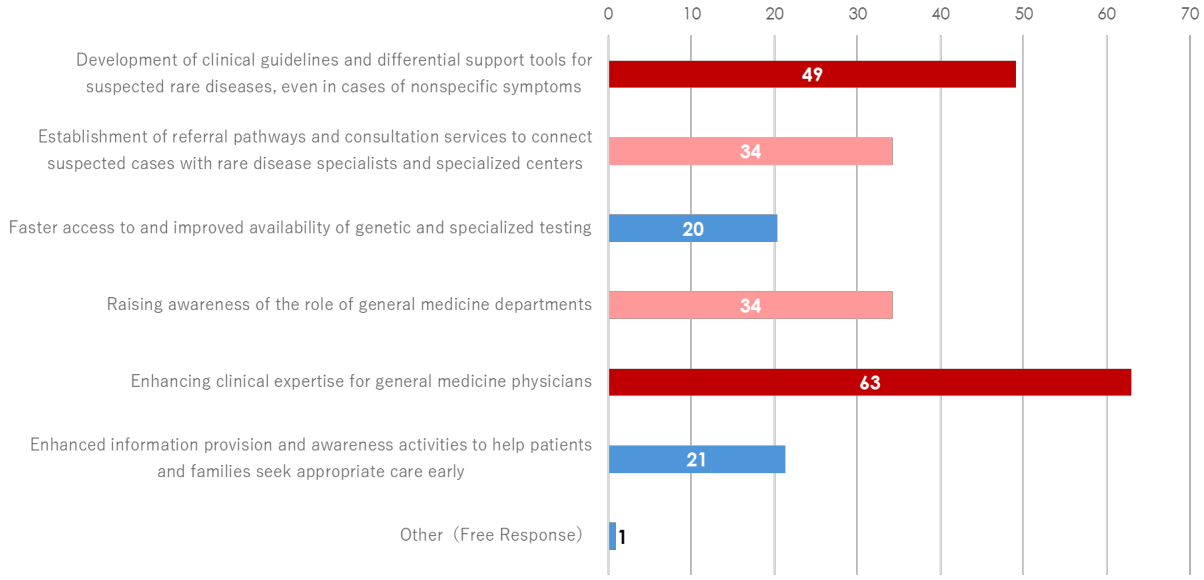
<Early Diagnosis Requires Both Enhancing Clinical Expertise and Support Systems for Differential Diagnosis>

Q. What types of support (up to three) are needed to promote early diagnosis of rare diseases? (Multiple answers allowed)



“ Enhancing clinical expertise for general medicine physicians” (63%) and “development of differential support tools and guidelines” (49%) ranked highest.

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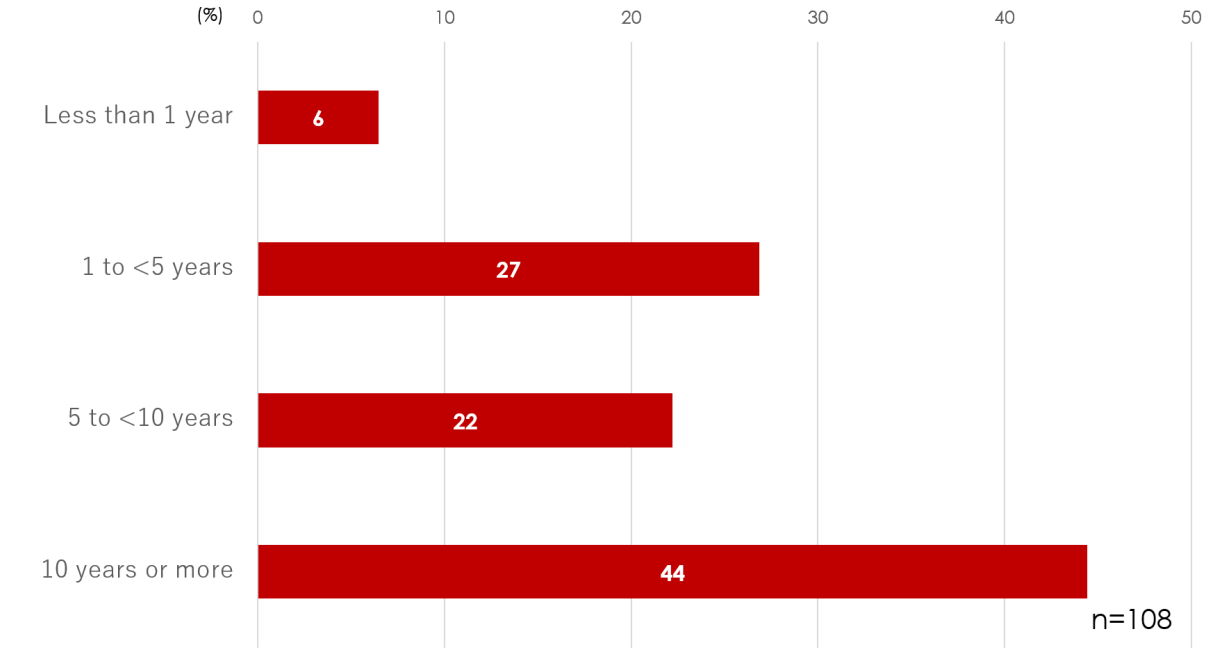
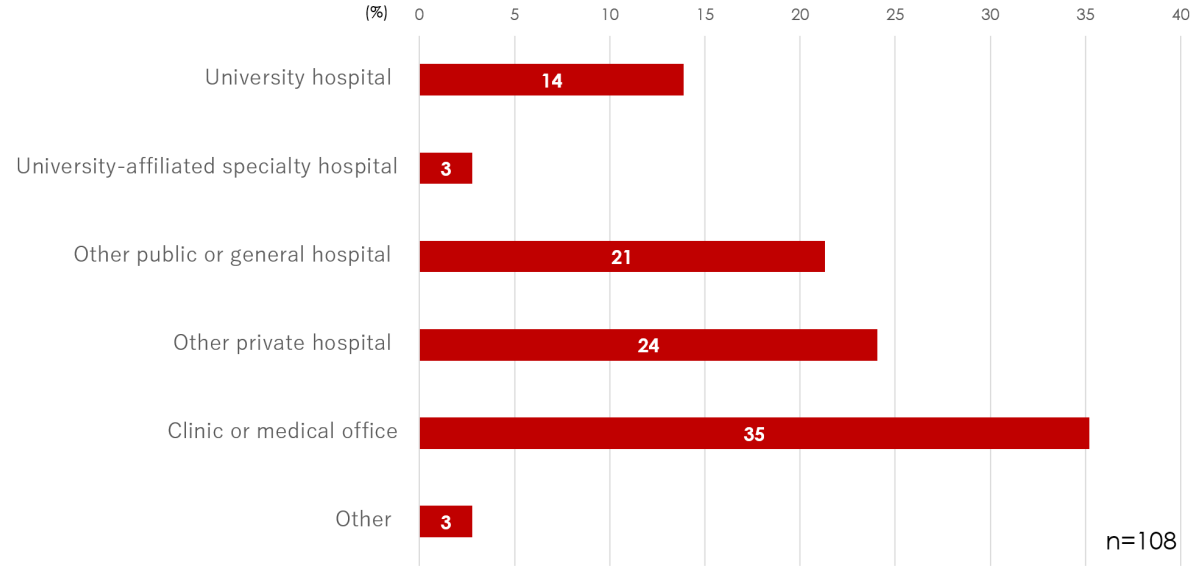
Respondent Demographics

Q. How many years of experience do you have in general medicine?



Q. What is the primary type of institution where you work?



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