DEFINITIVE HEALTHCARE
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2021 rare disease study

A Definitive Healthcare special report

A rare disease is a condition that affects fewer than 200,000 people in the United States.[1] There may be over 7,000 rare diseases affecting up to 30 million Americans.[2]

Following a previous study in 2019, Definitive Healthcare reached out to a group of healthcare providers to understand some of the challenges they face in diagnosing and treating patients with rare diseases.

Data was gathered in November and December 2021 via an online survey. We analyzed our ClaimsMx data to identify providers that had a diagnosis claim for one of over 142 rare diseases.[3] To qualify for the survey, the provider had to indicate experience diagnosing or treating rare diseases.

We collected responses from 150 individuals who work at about 128 healthcare organizations and hospitals. The top primary specialties of respondents include family practice, congenital/genetic disease, internal medicine, pediatric, neuroscience, oncology and radiology.

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More than one-third of respondents indicate that the top challenges providers face with treating rare disease today include a lack of rare disease education for physicians and a lack of awareness of symptoms related to rare diseases.

Nearly four out of 10 respondents think the greatest impact for providers addressing rare diseases today is collaboration with facilities and health systems specializing in rare disease.

About one-third of respondents highly rate their organization's ability to diagnose rare diseases. Meanwhile, just over a quarter of respondents highly rate their organization's ability to treat rare diseases.

Over 40% of respondents say that one of the ways to have the greatest impact in addressing rare diseases in the next five years is to increase physician education.

WE USED LATITUDE REPORTING TO ANALYZE

142	1.9M	633K	3+
Rare disease diagnosis codes	**Rare disease patients**	**Providers**	**Average patients with rare diseases providers diagnosing/treating**

Greatest challenges providers face around rare disease today

Respondents indicate that the lack of rare disease education for physicians (40%) and lack of awareness of symptoms related to rare diseases (39.3%) are the greatest challenges they face today around rare diseases.

In addition, shortages of physicians specializing in rare disease (38.7%), limited facilities and health systems specializing in rare diseases (38.7%) and a lack of clinical trials for many disease states (38.0%) contribute to the challenges providers face.

In the 2019 survey, providers indicated the top challenge they faced were the lack of clinical trials for many disease states followed by the lack of rare disease

education for physicians. The cost of genetic testing and reimbursement challenges rounded out the top four challenges identified in the 2019 study but have dropped slightly as a top concern based on the most recent survey.

WHAT DO YOU SEE AS THE GREATEST CHALLENGES AROUND RARE DISEASE IN HEALTHCARE TODAY?

40.0%
Lack of rare disease education

38.7%
Limited number of facilities/health systems specializing in rare disease states

39.3%
Lack of awareness of symptoms related to rare diseases

38.0%
Lack of clinical trials for many rare diseases

38.7%
Shortage of physicians specializing in rare disease states

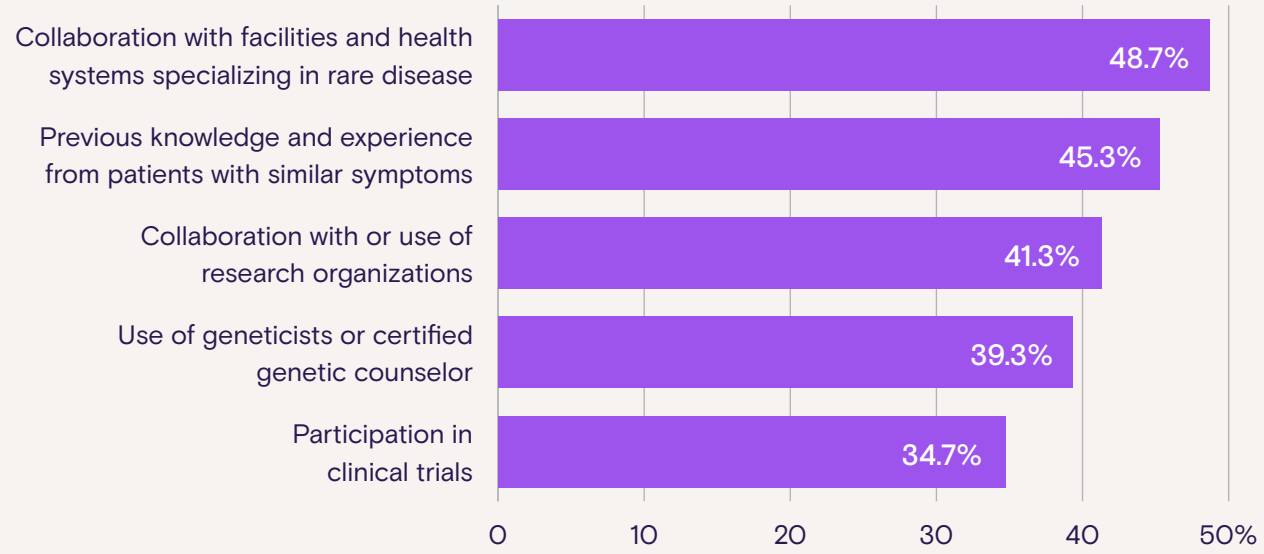
Greatest impact in addressing rare disease today

Respondents report collaboration with facilities and health systems specializing in rare disease (48.7%) will have the greatest positive impact in addressing rare diseases today.

In addition, previous knowledge and experience from patients with similar symptoms (45.3%) and collaboration with or use of research organizations such as Undiagnosed Disease Network [UDN] (41.3%) can help providers diagnose and treat patients with rare disease today.

Collaboration with facilities/health systems specializing in rare disease was also the top response from providers in 2019 survey, as well as participation in clinical trials.

WHICH OF THE FOLLOWING DO YOU FEEL HAS THE GREATEST POSITIVE IMPACT IN ADDRESSING RARE DISEASE IN HEALTHCARE TODAY?

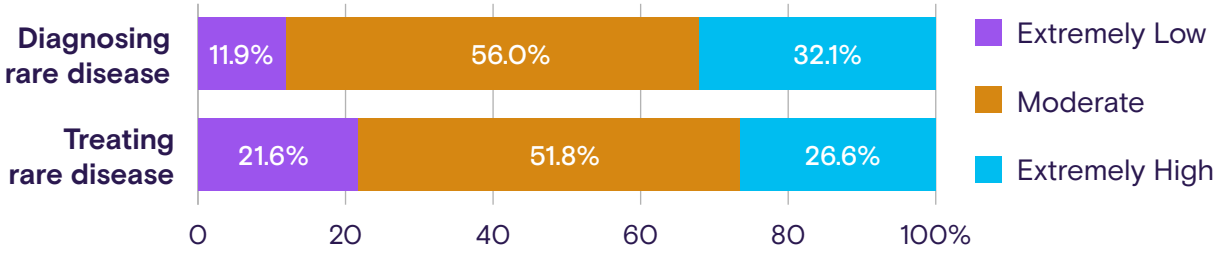


Organization's ability to diagnose and treat rare diseases today

About one-third of respondents highly rate their organization's ability to diagnose rare diseases. Out of a 10-point scale, the average rating is 7.03.

Over one-quarter of respondents highly rate their organization's ability to treat rare diseases. Out of a 10-point scale, the average rating is 6.53.

HOW WOULD YOU RATE YOUR ORGANIZATION'S ABILITY TO DIAGNOSE AND TREAT RARE DISEASE IN HEALTHCARE TODAY?



The 2019 survey asked for a rating for only the organization's ability to treat rare disease. The average rating out of 10 was 6.56 and less than a quarter of respondents highly rated their organization's ability to diagnose rare diseases.

Greatest impact in addressing rare disease in the next five years

Respondents state that increasing physician education (52%) and collaboration with facilities specializing in rare disease (44.7%) will have the greatest positive impact on rare diseases in the next five years.

In addition, next generation genomic sequencing has potential to help address diagnosing and treating rare disease in the future.

WHAT DO YOU FEEL WILL HAVE THE GREATEST POSITIVE IMPACT IN ADDRESSING RARE DISEASE IN HEALTHCARE WITHIN THE NEXT FIVE YEARS?

1	2	3
Increasing education for physicians	**Increasing collaboration with facilities/health systems specializing in rare disease**	**Next generation genomic sequencing**
52.0%	44.7%	41.3%

The top 3 responses from the 2019 survey were:

- → increased collaboration with facilities/health systems specializing in rare disease
- → expansion of/participation in clinical trials and
- → next generation genomic sequencing.

Key takeaways

Providers see areas of improvement in their organization's ability to diagnose and treat rare diseases

- Less than one-third of respondents rate their organizations as having a strong ability to diagnose rare diseases and about a quarter rate their organizations as having a strong ability treat rare diseases, indicating there is room for improvement in both areas.
- With advancement in genomic sequencing bringing costs down and making testing more accessible to patients, providers can better understand what conditions may be affecting patients and offer better relief and treatment.

To help better diagnose and treat patients with rare diseases, providers need expanded education on conditions, symptoms, patient journeys and therapeutic options

- Half of respondents think more education will have the greatest impact on addressing rare diseases in the next 5 years and 40% of respondents indicate it as a current challenge they face in their practice.
- Increased coordination between healthcare organizations and other members of the provider community can improve diagnosis and treatment of rare diseases, especially as 45% of respondents look to colleagues, geneticists and specialists for guidance on approaching rare diseases.

Study methodology

METHODOLOGY	
Data collection	Results gathered November through December 2021 via online survey
Inclusion criteria	Provider has diagnosis claim for one of 142 rare diseases; must indicate experience diagnosing or treating rare diseases
RESPONDENTS	
Total number	150 respondents from about 128 healthcare organizations and hospitals
Primary specialties	Family practice, congenital/genetic disease, internal medicine, pediatric, neuroscience, oncology and radiology

1 https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases

2 https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases

3 See appendix

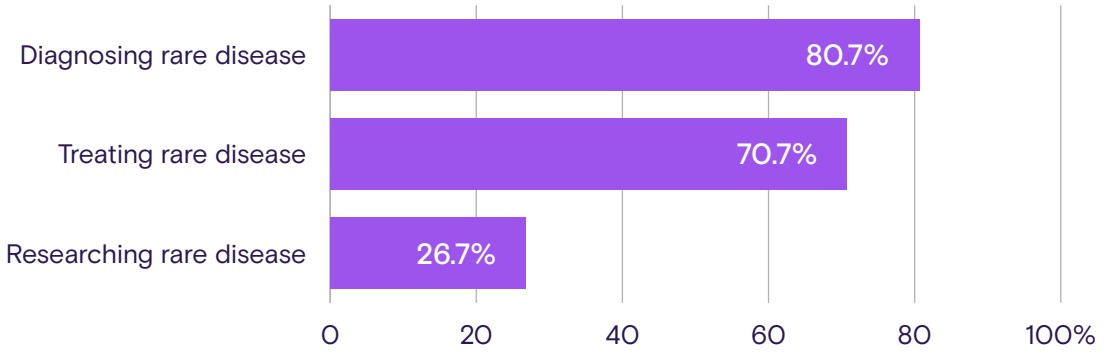
Appendix

LATITUDE REPORTING COHORT

Disease categories reviewed for provider claims

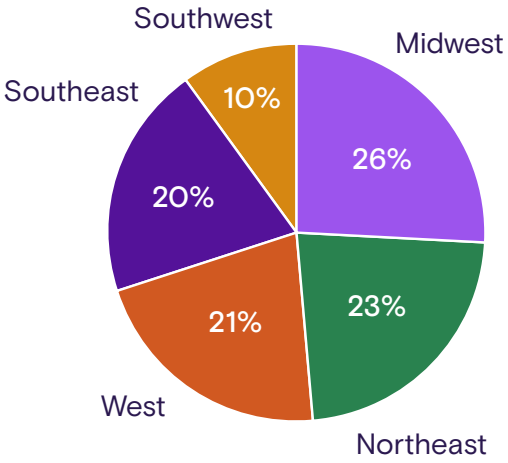
Achromatopsia CNGB3
Acromegaly
Amyloidosis
Amyotrophic Lateral Sclerosis
Angelman syndrome
Autoimmune Hemolytic Anemia
Cerebral adrenoleukodystrophy
Combined immunodeficiencies
Common variable immunodeficiency
Cushing's syndrome
Cystic fibrosis
Dense Deposit Disease
Duchenne Muscular Dystrophy
Ebola
Fabry Disease
Fragile X Syndrome
Gaucher disease
GI Stromal Tumor
Hemophilia
Huntington's Disease
Immunodeficiency associated with other major defects
Immunodeficiency with predominantly antibody defects
Mantle Cell Lymphoma
Morquio Syndrome
Multiple Sclerosis
Myasthenia gravis and other myoneural disorders
Myelodysplastic Syndrome
Narcolepsy
Neuromyelitis Optica
Morquio Syndrome
Multiple Sclerosis
Other coagulation defects
Pancreatic Cancer
Paroxysmal nocturnal hemoglobinuria
Primary Biliary Cholangitis
Retinal Dystrophy
Scleroderma
Spinal Muscular Atrophy
Tay-Sachs disease
Urea Cycle Disorders
Zika Virus

WHICH OF THE FOLLOWING BEST DESCRIBES YOUR ROLE/EXPERIENCE REGARDING RARE DISEASE IN HEALTHCARE TODAY?



TOP SPECIALTIES	%
Family Practice	12.0%
Congenital/Genetic Diseases	9.3%
Internal Medicine	8.0%
Pediatrics	8.0%
Neuroscience	7.3%
Oncology	7.3%
Radiology	7.3%
Hematology	5.3%
Pulmonary	5.3%

RESPONDENTS BY REGION



WHO ELSE DO YOU LOOK TO FOR GUIDANCE AND/OR WHAT OTHER METHODS DO YOU USE IN YOUR APPROACH TO RARE DISEASE TODAY?

45.3%
Colleagues, geneticists and specialists

10%
National / international organizations

10%
Other publications / literature

About Definitive Healthcare

At Definitive Healthcare, our passion is to transform data, analytics and expertise into healthcare commercial intelligence. We help clients uncover the right markets, opportunities and people, so they can shape tomorrow's healthcare industry. Our SaaS platform creates the path to commercial success in the healthcare market, so companies can identify where to go next.

Interested in discovering how our healthcare commercial intelligence can help grow your business? Contact your Definitive Healthcare Sales Executive to learn more, or start a free trial.

For more information, visit www.definitivehc.com.

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