RARE LIVER DISEASE SUMMIT 2022

Executive Summary

In June, 2022, the American Liver Foundation convened a two-day, virtual Rare Liver Disease Summit of leading liver disease experts and clinicians, patients, caregivers, advocacy organizations, and industry representatives to explore ways to improve health outcomes and overall patient experience for those living with a rare liver disease.

Through a series of presentations, panel discussions, and question-and-answer sessions, participants addressed several questions:

- How can people avoid misdiagnosis and/or achieve earlier diagnosis?
- How can we increase education about genetic testing, and how can we best address the stigmas associated with rare liver disease?
- How can telemedicine play a role?
- How can we reduce the patient burden and improve the overall patient experience?

From these interactive and informative sessions emerged several key strategies to help foster earlier and more accurate diagnosis of rare liver disease, raise the profile of genetic testing and clinical trial participation, and reduce perceived stigmas associated with liver disease. The strategies outlined within this Executive Summary reflect the general consensus of Summit participants as well as the patient voice. In addition, key themes and messages from the Summit reported here form the basis of a newly published Rare Liver Disease Patient Bill of Rights to help patients and families develop better working relationships with their health care teams. The ALF Rare Liver Disease Summit was organized in conjunction with the Liver Diseases Branch of the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) of the National Institutes of Health (NIH).

About Rare Liver Diseases

In the United States a disease is considered rare if it affects fewer than 200,000 people. Collectively 30 million Americans are currently living with a rare liver disease. There are more than 7,000 types of rare liver diseases. Some of the more common ones include Acute Hepatic Porphyria, Alagille Syndrome, Alpha-1 Antitrypsin Deficiency, Biliary Atresia, Crigler-Najjar Syndrome, Galactosemia, Glycogen Storage Disease, Lysosomal Acid Lipase Deficiency, Primary Biliary Cholangitis, Primary Sclerosing Cholangitis, Progressive Familial Intrahepatic Cholestasis (PFIC), and Wilson Disease.

Key Themes and Messages from the 2022 Rare Liver Disease Summit

Quality of life issues that matter most to patients

- Many patients with rare liver diseases report feeling alone, overwhelmed, and misunderstood by others.
- The average time to receive a diagnosis of a rare liver disease is five years, and some patients may never receive a definitive one. Living with prolonged uncertainty commonly causes feelings of frustration, isolation, loneliness, anger, depression, and anxiety.
- There is social stigma associated with rare liver diseases. Patients feel misunderstood by others who mistakenly think their liver disease is a result of alcohol or drug use. Some people avoid or delay seeking treatment due to fear that they will be disbelieved by their doctors, family, friends and others.

Avoiding misdiagnosis/achieving earlier diagnosis

- Rare liver diseases are complicated to diagnose and treat and are challenging to patients, caregivers, and health care providers alike. However, every patient ,deserves the highest level of care from a trusted medical team who listen to and address their questions and concerns.
- Health care teams are multidisciplinary, spanning medical and surgical specialties such as hepatology, gastroenterology, cardiology, pulmonology, and internal medicine/primary care. Coordinating care plans among multiple specialists can seem overwhelming to patients and challenging for clinicians, but it is essential for the best health outcomes.
- There is low awareness of rare liver diseases among the medical community at large. It is not uncommon for patients to hear, “You are the first patient I’ve ever seen with these symptoms.” This often leads to delayed diagnosis or misdiagnosis, and patient loss of trust in the care team. Emphasizing liver function, health, and disease in medical education and training of healthcare professionals should be a priority.
- In the United States, there are several academic medical centers of excellence in the diagnosis and treatment of rare liver diseases, many offering clinical trials of new therapies and treatment approaches. Summit participants unanimously agreed upon the importance of visiting a center of excellence to receive care, and the value of participation in clinical trials, both to the patient who may benefit and to researchers who gain knowledge about these rare diseases. The NIH maintains a searchable, public database of all clinical trials at https://clinicaltrials.gov/.

The patient’s voice is vital in improving health outcomes

- Patients should feel empowered to actively participate in their care plan. This means selecting and engaging with a health care team that is open, available, and willing to listen to and learn along with them, throughout their entire health journey.

- Summit participants – patients, caregivers, clinicians and advocates – made several recommendations for actively participating in one's own care, including: bringing copies of tests results to all clinic visits; keeping a journal to document day-to-day physical and mental health symptoms for discussion with the health care team; and bringing a caregiver along for all visits to help absorb the medical information.

Telemedicine and genetic testing

- Telemedicine can improve access to care and may even be essential for patients who live far from a center of excellence, have inadequate transportation alternatives, or who have compromised immune systems. Telehealth can also expand the reach of clinical trial participation. However, telemedicine should not replace in-person visits. Physical exams are extremely important to accurately assess a patient's health (omit comma after "health") and maintain productive working relationships among patients, caregivers and health care providers.
- Genetic testing can often, but not always, confirm a rare liver disease diagnosis. By shedding light on the genetic drivers of an individual patient's disease, it can guide treatment decisions and clinical trial participation by helping clinicians predict whether a patient may or may not respond to new molecular targeted therapies. Some patients avoid genetic testing out of fear of negatively impacting family relationships and/or problems with health insurance.
- With rare diseases, there is the added burden of insurance companies not paying for genetic testing or treatments. This can significantly delay diagnosis. By forming coalitions and collaborations, patient advocacy groups can educate insurers and policymakers and help influence health care legislation.

Education and advocacy are key to improving outcomes and reducing stigma

- There are numerous patient advocacy groups and nonprofit foundations that raise funds for rare liver disease research, offer support services and educational events for both patients and caregivers, and educate elected officials who can influence health care policy and research funding. By joining together in coalitions and participating in events such as the ALF Rare Liver Disease Summit, advocates amplify the patient's voice, educate others, and raise awareness about rare liver disease. Education leads to understanding and dispels stigma.
- Patients and caregivers are encouraged to talk openly about their rare liver diseases. It is important for researchers, clinicians, insurers, elected officials, and the public at large to hear personal stories of what it is like to live with a rare liver disease.

The Bottom Line – Strategies and Solutions for Living Well With Rare Liver Disease

- Patients must be their own best advocates for their health. This means actively engaging in your own care, surrounding yourself with a team of health care providers who listen to you and are willing to learn alongside you, and sharing with them your symptoms, questions and concerns.

- Trust your instincts and ask for a second opinion concerning your diagnosis and treatment plan if you feel misunderstood or disbelieved by your health care provider.
- Know that there is strength in numbers, and you are not alone. Build your team of supporters including family, friends, and health care professionals. Engage with other rare liver disease patients and caregivers for emotional support.
- Educate others about rare liver disease to reduce misinformation and stigma. Connect with rare disease patient groups and advocacy organizations to share your personal story with elected officials and help inform public policy.

Additional Resources

1. American Liver Foundation
2. National Institute of Diabetes and Digestive and Kidney Disease – Liver Branch
3. NIH Clinical Trials Database
4. National Organization for Rare Disorders (NORD)
5. LiverTox© - The NIH maintains this unbiased and searchable database of information about liver injury attributable to prescription and nonprescription medications as well as herbal and dietary supplements.
6. Rare Genomes Project at The Eli and Edythe L. Broad Institute of MIT and Harvard

ALF Rare Live Disease Summit Speakers

Theo Heller, MD
Chief of the Translational Hepatology Section, National Institutes of Health

Shannon Palmatier
Patient Advocate for Progressive Familial Intrahepatic Cholestasis (PFIC)

Bruce Dimmig
Patient Advocate for Nodular Regenerative Hyperplasia and NASH

Kim Crowell
Patient Advocate for Portopulmonary Hypertension and Liver Transplant

Barb Pitts
Patient Advocate with Undiagnosed Rare Liver Disease

Nehna Abdul Majeed, MD
Clinical Fellow, Hepatology Section, National Institutes of Health

ALF Rare Live Disease Summit Speakers, Continued

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Clinical Fellow, Hepatology Section, National Institutes of Health

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Joanne Hatchett
Medical Science Liaison, PSC Partners Seeking a Cure

Scott Santarella
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American Liver Foundation Staff in Attendance

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