PBC and Beyond: How Technology is Shaping the Treatment of Rare Liver Diseases

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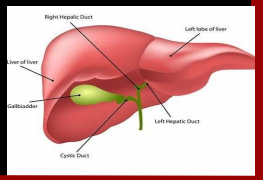
The Liver

The liver is a vital organ located in the upper right quadrant of the abdomen, beneath the diaphragm. It plays a crucial role in various bodily functions, including metabolism, detoxification, and digestion. One of its primary responsibilities is processing nutrients absorbed from the digestive tract, converting them into essential components such as glucose, proteins, and fats. Additionally, the liver detoxifies harmful substances by metabolizing drugs and neutralizing toxins, ensuring they are safely eliminated from the body.

Healthy Liver →



The Impact of Technology

Technology is having a profound impact on the diagnosis, treatment, and management of rare liver diseases such as Primary Biliary Cholangitis (PBC). Advances in medical imaging, such as magnetic resonance elastography (MRE) and liver biopsy alternatives, allow for earlier and more accurate assessments of liver damage, reducing the need for invasive procedures. Additionally, genetic testing and biomarker research are enhancing diagnostic capabilities by identifying new indicators for PBC, leading to faster and more precise diagnoses. The use of telemedicine is also growing, enabling patients to receive consultations and ongoing care from specialists without needing to travel long distances, which is particularly valuable for individuals in underserved areas.

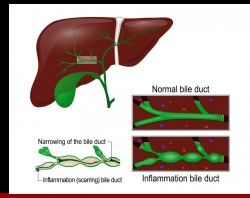
In treatment, technology is revolutionizing drug development and personalized medicine. Innovations in artificial intelligence (AI) and machine learning are being used to analyze large datasets to identify new drug candidates and predict patient responses to specific treatments. For example, AI models are accelerating the discovery of compounds that could slow or halt the progression of PBC. Digital health tools, such as mobile apps and wearable devices, are also helping patients track their symptoms, medication adherence, and liver function, providing real-time data that healthcare providers can use to adjust treatments more effectively. These technological advancements are transforming how PBC is diagnosed and treated, offering hope for more effective therapies and improved patient outcomes.

Primary Biliary Cholangitis (PBC)

Primary Biliary Cholangitis (PBC) is a chronic autoimmune liver disease that gradually destroys the small bile ducts, leading to bile accumulation, liver inflammation, and eventual cirrhosis or liver failure. The disease primarily affects middle-aged women and is often associated with other autoimmune conditions. Diagnosis is typically based on blood tests detecting anti-mitochondrial antibodies (AMA) and elevated liver enzymes, particularly alkaline phosphatase (ALP). In unclear cases, a liver biopsy may be performed to confirm bile duct damage.

PBC often presents with fatigue, itching (pruritus), and dry eyes or mouth, though some patients remain asymptomatic for years. As the disease progresses, symptoms like jaundice, darkened skin, and cholesterol deposits around the eyes may appear. The impaired bile flow affects digestion, leading to vitamin deficiencies and metabolic issues. Without treatment, prolonged inflammation and scarring can cause severe liver dysfunction, making early diagnosis and intervention crucial to slowing disease progression and improving quality of life.

Liver with PBC →



Policies

FDA Restrictions on Ocaliva (Obeticholic Acid):
In 2021, the U.S. Food and Drug Administration (FDA) restricted the use of Ocaliva for PBC patients with advanced cirrhosis due to risks of serious liver injury. Despite these restrictions, reports indicated that some patients with advanced cirrhosis continued using the medication, leading the FDA to issue further warnings in 2024. Healthcare providers are now advised to carefully assess liver disease severity before prescribing Ocaliva and to monitor patients regularly for potential liver-related side effects.

AASLD Practice Guidance on PBC:
The American Association for the Study of Liver Diseases (AASLD) released updated practice guidance in 2021 for the diagnosis and treatment of PBC. This guidance emphasizes early diagnosis, the use of ursodeoxycholic acid (UDCA) as first-line therapy, and consideration of obeticholic acid for patients not responding adequately to UDCA. The guidelines also highlight the importance of monitoring for disease progression and managing associated symptoms to improve patient outcomes.

← Summarized Info to take mobile

Treatments

Primary Biliary Cholangitis (PBC) is among the approximately 5% of rare diseases that have an FDA-approved treatment. The first-line therapy for PBC is Ursodeoxycholic Acid (UDCA), administered at a dose of 13–15 mg/kg/day. UDCA has been shown to improve survival free of liver transplantation. For patients who do not respond adequately to UDCA,

Obeticholic Acid (OCA) is available as a second-line treatment. Patient perspectives on these treatments vary; while some report significant improvements in symptoms and quality of life, others experience side effects or insufficient symptom relief, underscoring the need for personalized treatment approaches.

New drugs are now approved for second line therapy including Elafibranor and Seladelpar.

Mental Integrity

Living with a rare disease like Primary Biliary Cholangitis (PBC) can lead to significant mental and emotional challenges, including anxiety, depression, and feelings of isolation due to the chronic nature of the illness and its impact on daily life. Patients may experience "illness uncertainty," struggling with the unpredictability of symptoms and the long-term outlook of their condition. Support groups, counseling, and mental health services can play a crucial role in helping individuals cope. Connecting with others through online forums or local support networks provides emotional support and practical advice. Additionally, incorporating mindfulness practices, such as meditation or yoga, may reduce stress and improve overall well-being. Healthcare providers should assess mental health regularly and refer patients to appropriate resources to address psychological needs, fostering a holistic approach to managing PBC.

PBC Research Foundation

The PBC Research Foundation (PRF) is a patient-driven initiative founded by Dr. Cecilia Dueñas focused on collecting and managing vital health data from individuals with Primary Biliary Cholangitis (PBC), a rare liver disease. Through its secure, patient-owned registry, the foundation ensures that data is confidential, compliant, and regulatory, while amplifying the patient voice in both current treatment and future research. This registry not only supports the development of new treatments for PBC but also serves to enhance existing therapies by providing real-world, unbiased data and biospecimens. Importantly, it seeks to collect data from all PBC patients, not just those involved in clinical studies, to create a comprehensive understanding of the disease.

The mission of the PRF is to accelerate clinical breakthroughs in rare liver diseases by harnessing patient-centered data. By collaborating with researchers, clinicians, and regulatory bodies, the foundation aims to bridge the gap between patient experiences and scientific advancements.

Survey →

Resources: Mayoclinic.org, PBCresearchfoundation.org, Liverfoundation.org, AASLD.org