

Gene Chip to Diagnose Genetic Mutations Related to Liver Disease

TECHNICAL FIELD

Diagnostic: Liver Disease (2005-0306)

BACKGROUND

In 2004, there were 5,000 liver transplants in adults and 500 in children. Fifty-three percent of pediatric liver transplantation is due to cholestasis of the liver. In children ages 6-11, 1 in 500 will develop a form of liver disease, and in children ages 12-19, 1 in 250 children will develop a form of liver disease. Neonatal severe liver disease occurs in about 1 in 2,500 live births with cholestasis being a main cause.

The broad array of causes of cholestasis (genetic, metabolic, inflammatory, and drug- or toxin-induced disorders) creates a challenge for physicians to diagnose a specific disease. Therefore, the treatment of affected children is not disease-specific or aimed at optimizing care to help reduce liver transplantations. Due to the lack of effective medical therapies, liver transplantation has become the standard treatment for endstage, as well as fulminant, liver disease in children. A considerable number of adult liver disease cases begin in childhood. Some of the liver diseases of adults (e.g. gallstones, cholestasis of pregnancy) share genetic basis with some of the liver diseases in children. Chronic liver disease is one of the most expensive digestive diseases, costing roughly \$1.6 billion annually.

The availability of a single test that diagnoses the genetic mutations of liver disease will facilitate diagnostic algorithms and implementation of specific treatments.



TECHNOLOGY

There is a significant unmet need for a non-invasive means for accurate diagnosis, staging, and grading of liver diseases. Early diagnosis will help to elucidate the causes of these diseases and develop practical means of treatment and prevention. Dr. Jorge Bezerra at Cincinnati Children's Research Foundation (CCRF), has assembled a diagnostic gene chip that can be used in early diagnosis of liver disorders. Termed the "JAUNDICE CHIP", this technology is a re-sequencing chip that allows for full DNA sequencing of specifically identified genes involved in liver disease. Furthermore, the detection of mutations will be possible by hybridizing these chips with DNA isolated from blood samples from patients with liver disease.

This "JAUNDICE CHIP" fills a technology gap and a clinical need by offering a unique opportunity to simultaneously screen for mutations in the most common genes causing pathologic jaundice and other forms of liver disease in children and adults.

APPLICATIONS

1. Gene-based test for liver disease diagnosis and prognosis
2. Research tool

ADVANTAGES

- Rapid, accurate test
- Standardized diagnostic tool
- Early intervention and better disease management
- Potential for savings to health care system

INVESTIGATOR

Jorge Bezerra, MD, PhD
Professor
Division of Gastroenterology, Hepatology and Nutrition
Cincinnati Children's Hospital Medical Center

STATUS

Patent applications pending.

CONTACT

Korie Counts, PhD
Technology Manager
korie.counts@cchmc.org
513-636-6736