CLINICAL AND LABORATORY PROFILE OF CHILDREN WITH PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS ATTENDING LIVER CLINIC IN ALEXANDRIA UNIVERSITY CHILDREN'S HOSPITAL

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Introduction

Progressive familial intrahepatic cholestasis (PFIC) is a heterogeneous group of rare autosomal recessive liver disorders of childhood characterized by mutations in genes encoding proteins involved in the hepatocellular transport system. Although the prevalence is unknown, its estimated incidence ranges between 1 in 50,000 and 1 in 100,000 live births.

Most patients diagnosed with PFIC present with cholestasis, which is marked by jaundice and pruritus. Other findings include inadequate weight gain, feeding difficulties, and hepatosplenomegaly.

Liver biopsy can diagnose the disease but Genetic testing is the gold standard.

Treatment for patients with PFIC involves both medical and surgical approaches. Medical management includes dietary modifications, vitamins, and medications, while surgical treatment involves biliary diversion and liver transplantation.

Aim of the Work

The study aimed to describe the clinical, laboratory, histopathological and genetic profiles of children with PFIC Attending the Liver Clinic at Alexandria University Children's Hospital.

Subjects and Methods

The study included all children diagnosed as PFIC by liver biopsy or genetic testing attending the Liver Clinic at Alexandria University Children's Hospital (22 patients) from June 2017 to July 2024.

It's a retrospective study with a prospective aspect. Data was obtained from the patients and their admission files and the patients were evaluated for:

I. History taking with special emphasis on:
Age, Sex, Residence, Age of presentation, Consanguinity and The presenting symptoms.

II. Complete physical examination on admission and follow up.

III. Laboratory invesigations: CBC, ALT, AST, TSB, DSB, GGT, ALP, serum albumin, blood glucose and bleeding profile.

IV. Abdominal Ultrasonoghraphy

V. Liver biopsy

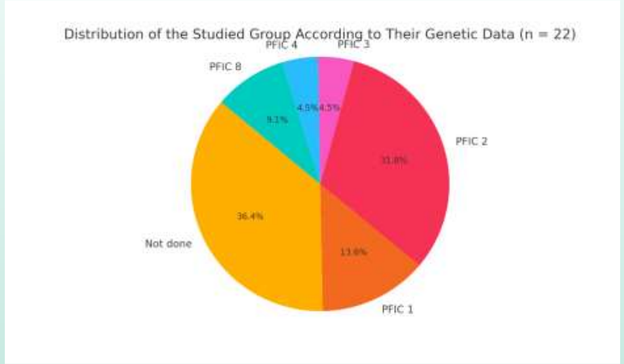
VI. Molecular analysis (if available)

B. Follow up: Patients were evaluated regularly in the Liver Clinic.

Results

Table: Patients outcomes categorized by different treatment interventions:(n = 22)

	No.	%
Improving		
Yes	8	36.4
Medical Rx	3	13.6
Medical Rx and biliary diversion	4	18.2
Liver transplant	1	4.5
Complications	14	63.6
P.HTN	14	63.6
ESLD	13	59.1
SBP	1	4.5
Mortality		
Death	6	27.3
Survival	16	72.7



Figure

Conclusion

PFIC is a rare and severe condition. Jaundice is the most frequently observed symptom, while common complications include growth retardation, vitamin deficiencies, portal hypertension, and end-stage liver disease. Pruritus, which affects the majority of cases, is the most troubling symptom and often prompts surgical interventions like biliary diversion or liver transplantation.

Biliary diversion can alleviate pruritus and slow disease progression in most cases, but liver transplantation remains the only definitive cure.