GUT MICROBIOME AND MYCOBIOME ALTERATIONS IN PAEDIATRIC PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS

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Paediatric progressive familial intrahepatic cholestasis is associated with consistent microbial dysbiosis characterized by reduced bacterial diversity, decreased beneficial taxa, and increased Candida abundance across all patient groups.

KEY POINTS

- Microbial diversity measures including species richness, Shannon diversity, and absolute bacterial cell counts were consistently reduced in all PFIC patient groups compared with healthy controls, with multivariate analysis confirming significant differences ($p = 0.001$).
- Faecalibacterium was consistently reduced across all patient groups relative to controls, while Candida levels were increased across all PFIC groups with highest abundance in untreated patients.
- IBAT inhibitor therapy was associated with neither improvement nor worsening of dysbiotic changes in the microbiome.
- Age emerged as a major confounding factor, with 21% of the cohort younger than 1.5 years showing distinct microbial patterns and requiring exclusion from comparative analyses.
- The findings support a role of the gut-liver axis in PFIC, though the cohort included 75 paediatric participants (45 PFIC patients and 30 healthy controls) from this rare disease affecting approximately 1 in 50,000 children.

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