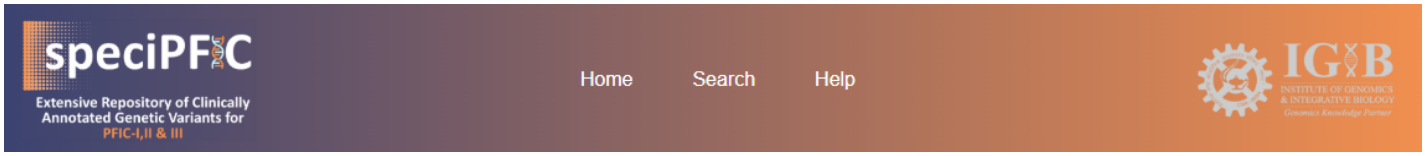

USER MANUAL

About speciPFIC

speciPFIC is a curated database for clinicians and geneticists studying Progressive Familial Intrahepatic Cholestasis (PFIC I–III). It compiles variants in the three key genes—ATP8B1, ABCB11, and ABCB4—systematically annotated from ClinVar, LOVD, Google Scholar, and other resources.

The platform includes four main tabs—**'Home'**, **'Search'**, **'Contact'**, and **'Help'**—to support easy navigation, enhanced data accessibility, and an improved user experience.

Home page

The **Home** page provides a brief overview of **speciPFIC**, highlighting its purpose, contents, and clinical significance. It features a graphical abstract summarizing the database structure, data sources, and key findings. Additionally, pie charts display the Functional Classification and ACMG Classification of variants in the three genes (ABCB4, ABCB11, and ATP8B1). The page also includes buttons for accessing the contact section and user manual.

Search page

The **Search** page allows users to query for speciPFIC through a search bar and a selection box to choose any of the three genes (ABCB4, ABCB11, and ATP8B1). Results are displayed on the page with detailed information about the location and classification of each genetic variant.

Users can search using the following formats:

- **Variant format**: chr-position-reference allele-alternate allele (e.g., 21-38123372-T-C or 21:38123925:C:T)
- **Amino acid change (AAChange)**: e.g., S658G, P656S, R565Q
- **ACMG classification**: e.g., Benign, Pathogenic, Likely Pathogenic, VUS
- **dbSNP ID**: e.g.,rs754770911, rs1160711086

If the variant or corresponding information exists in the database, matching results will appear under the Search Results section, below the “Example Search” window. Users can control the number of entries displayed using a dropdown menu.
Search results are presented in a table containing:

- **Gene name**
- **AAChange**
- **dbSNP ID**
- **Mutation Effect**
- **Population**
- **ACMG**

If multiple results are returned, users can refine their search using chromosomal position, dbSNP ID, or ACMG using a dropdown menu.The last column in each row is labeled **More Info**, which contains a button. When clicked, a popup window appears displaying detailed information about the selected variant. If the database does not have the variant, it will show “There are no matching data within the database”.

Contact page
The **Contact** page provides essential contact information for users seeking assistance or further details about **speciPFIC**. It includes the institutional address of CSIR-Institute of Genomics and Integrative Biology (CSIR-IGIB), Mall Road, New Delhi, India. Key contacts listed are Dr. Binukumar BK, Principal Scientist , and Mr. Mohammed Swalih KT, Database Developer. Users are encouraged to reach out for queries, feedback, or collaboration opportunities related to the database.

Help page
The **Help** page is designed to guide users in understanding and utilizing **speciPFIC**. It includes three informative sections:

- **About PFIC Disease** – A brief summary of Holocarboxylase Synthetase Deficiency with a link to an external resource for more information.
- **About Genes** – Details about the ABCB4, ABCB11, and ATP8B1 genes, each with a link to their respective OMIM pages for in-depth genetic insights.
- **User Manual** – Access to the full user manual offering detailed instructions on navigating and using the database.

Additionally, a link button to the BK Lab website is provided at the bottom for users interested in learning more about the lab's research and other projects.

Additional Feature

On the left side of every page, the official CSIR-IGIB logo is prominently displayed. Clicking this symbol redirects users to the CSIR-IGIB official website for institutional information and additional resources.

speciPFIC database is created and maintained by CSIR-Institute of Genomics and Integrative Biology (CSIR-IGIB).