Database and web server

We developed a web-based database for the exploration of Alagille syndrome and related data incorporating clinical, and genetic information from previous studies. The website was hosted on a live server using Apache, with MariaDB as the database management system, which is a fork of MySQL. The collected genetic mutation data, clinical symptoms, and patient records were stored in MariaDB with indexing applied to optimize the search queries and improve efficiency.

The front end of the database was made using HTML5 for structure, CSS3 for styling and JavaScript to handle front-end operations like form submissions, sorting and filtering of data, and animations. PHP was used to handle server-side logic and connect the application to MariaDB. The CRUD operations, form submissions, and search queries were handled by PHP. A dedicated search page was created to help users navigate the database efficiently. The search interface used an HTML form, with JavaScript dynamically handling and displaying the results.

Database Interface and Functionalities

The Home page of the database was designed to be user-friendly and intuitive, allowing seamless navigation for researchers and clinicians. Organized tabs were made to facilitate browsing through mutations of JAG1 and NOTCH2 genes. A dedicated search page facilitated users to efficiently query the entire database with suitable keywords.

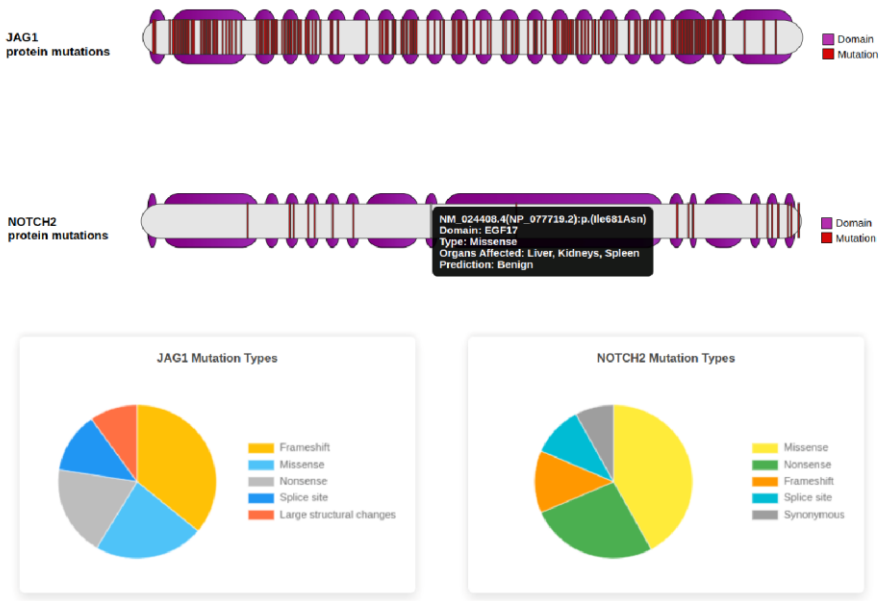

Figure1: Interactive visualizations on the home page

Interactive graphical representations were added to the home page that represented JAG1, and NOTCH2 proteins, their domain structures, and the mutations that were reported in each domain. Clicking on a mutation automatically searched and provided more details on the mutation from the database. Additionally, an interactive pie chart was added to represent different mutation types and illustrate their frequency for each gene. Clicking on a mutation category redirected the user to a larger mutation table, providing detailed information on the selected mutation type.

Browsing and Data Exploration

We made a structured browsing system for efficient exploration of the database through a hierarchical drop-down menu. The browse tab, on click, provided access to JAG1 and NOTCH2 mutations, with JAG1 further expanding to subcategories based on mutation types: Missense, nonsense, Frame-shift, Large Structural changes, and Splice-site variants. This approach enabled users to quickly locate and examine specific mutation datasets without needing to perform a manual search.

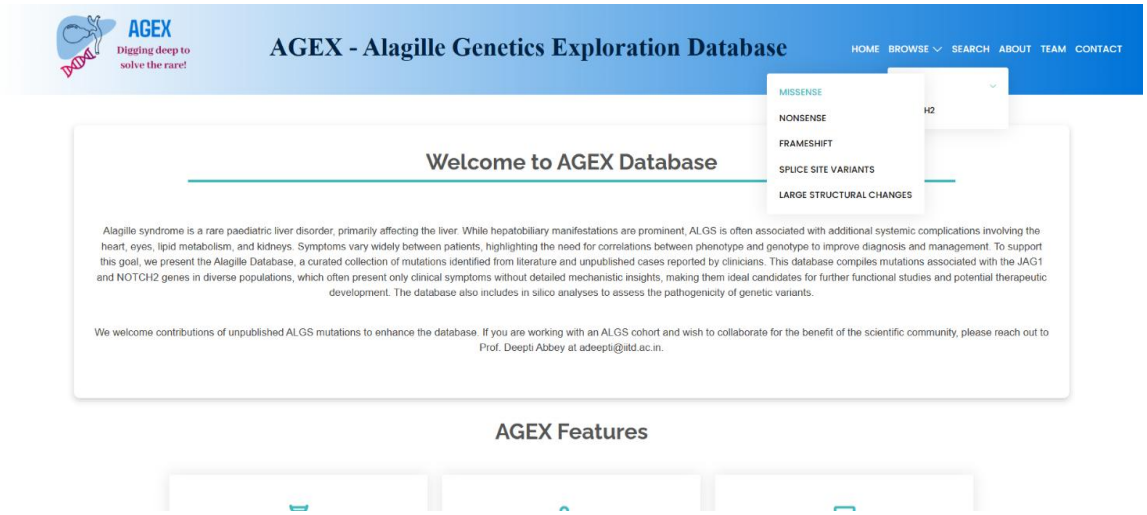

Figure2: Browsing and Data exploration options on database

Advanced Search Functionalities

The dedicated search page offered users a comprehensive querying system that allowed filtering data based on desired criteria. The criteria for the search included gene name, mutation type, ethnicity, gender, and other fields available in the database. The search function retrieved and displayed results across all mutation categories, providing maximum information efficiently.

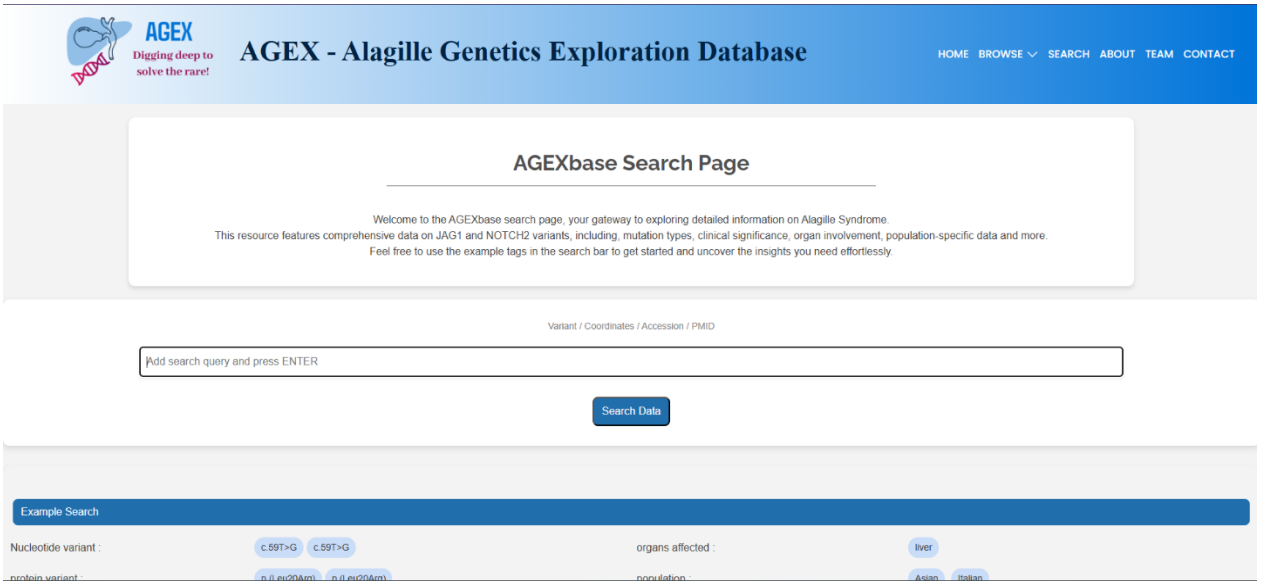

Figure3: Dedicated search page of the database

When search results yielded large datasets, an additional search tab appeared above each table, for searching within results. This allowed users to refine their queries within each table, offering greater flexibility in filtering and pinpointing relevant information to further enhance usability. This ensures that even for extensive datasets, users could efficiently extract the most relevant data without the need to repeat their search.

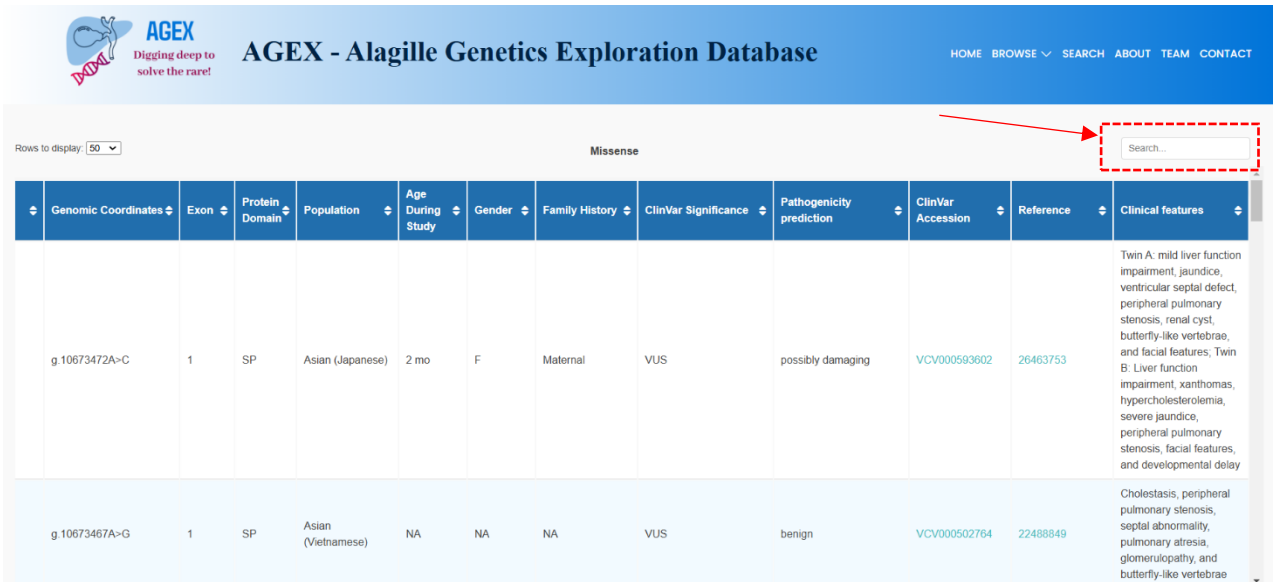

Figure4: Example results for a query on the search page along with search bars within results