The Kidney-Genetics Documentation

Bernt Popp, Nina Rank, Constantin Wolff, Jan Halbritter

2023-05-23

Contents

Preface

This documentation is intended to describe the Kidney-Genetics[1] project.

[1] https://github.com/halbritter-lab/kidney-genetics

Objective

How can we address the lack of a unified and standardized database of kidney disease-associated genes, which hampers diagnosis, treatment, and research comparability in the field of kidney diseases?

Methods

To create a comprehensive and standardized database of kidney-related genes, we employed the following methods:

1. Utilized data from Genomics England and Australia PanelApp.
2. Conducted a comprehensive literature review of published gene lists.
3. Collected information from clinical diagnostic panels for kidney disease.
4. Performed a Human Phenotype Ontology (HPO)-based search in rare disease databases (OMIM, Orphanet).
5. Employed a PubTator API-based automated literature extraction from PubMed.

We also developed an evidence-scoring system to differentiate highly confirmed disease genes from candidate genes.

0.1 Results

The "Kidney-Genetics" database currently includes detailed information on 2,906 kidney-associated genes. Notably, 439 genes (15.1%) are present in three or more of the analyzed information sources, indicating high confidence and their potential for diagnostic use.

To ensure currency, Kidney-Genetics will be regularly and automatically updated. We will also provide phenotypic and functional clustering results to facilitate gene grouping.

0.2 Conclusion

Kidney-Genetics is a comprehensive and freely accessible database that researchers can use to analyze genomic data related to kidney diseases. The database is regularly updated through a standardized pipeline and an automated system, ensuring it remains up-to-date with the latest advancements in kidney research and diagnostics.

By utilizing Kidney-Genetics, clinicians and researchers can enhance their understanding of the genetic aspects of kidney disorders.

0.3 Outlook

Future goals include manual curation and the assignment of diagnostic genes to specific nephrological disease groups, such as syndromic vs. isolated, adult- vs. pediatric-onset, and cystic vs. nephrotic, among others.

1 Analyses result tables

1.1 Main table: Merged analyses sources

This table shows the merged results of all analyses files as a wide table with summarized information.

approved_symbol	hgnc_id	evidence_count	list_count	01_PanelApp	02_Literature	03_DiagnosticPanels	04_HPO	05_PubTator
All	All	All	All	All	All	All	All	All

1.2 Result table: PanelApp

This table shows results of the first analysis searching kidney disease associated genes from the PanelApp project in the UK and Australia.

approved_symbol	hgnc_id	gene_name_reported	source	source_count	source_evidence
All	All	All	All	All	All

1.3 Result table: Literature

This table shows results of the second analysis searching kidney disease associated genes from various publications.

approved_symbol	hgnc_id	gene_name_reported	source	source_count	source_evidence
All	All	All	All	All	All

1.4 Result table: Diagnostic panels

This table shows results of the third analysis searching kidney disease associated genes from clinical diagnostic panels for kidney disease.

approved_symbol	hgnc_id	gene_name_reported	source	source_count	source_evidence
All	All	All	All	All	All

1.5 Result table: HPO in rare disease databases

This table shows results of the fourth analysis searching kidney disease associated genes from a Human Phenotype Ontology (HPO)-based search in rare disease databases (OMIM, Orphanet).

approved_symbol	hgnc_id	gene_name_reported	source	source_count	source_evidence
All	All	All	All	All	All

1.6 Result table: PubTator

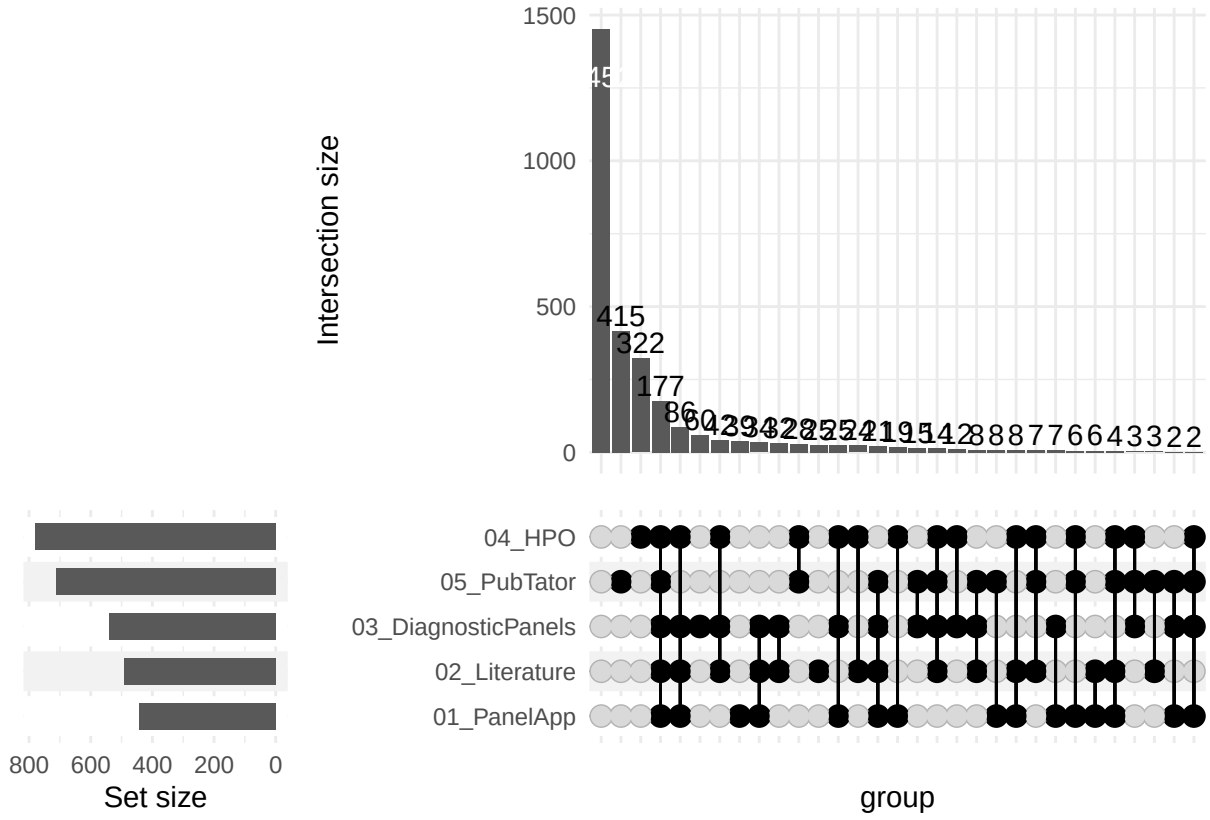
This table shows results of the fifth analysis searching kidney disease associated genes from a PubTator API-based automated literature extraction from PubMed.

approved_symbol	hgnc_id	gene_name_reported	source	source_count	source_evidence
All	All	All	All	All	All

2 Analyses plots

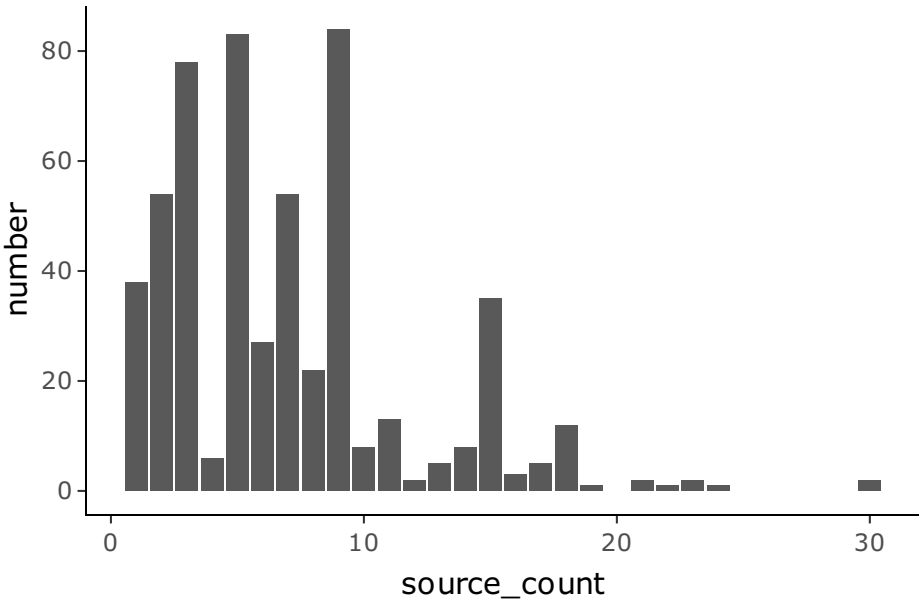
2.1 UpSet plot of merged analyses sources

UpSet plot of the merged analyses.



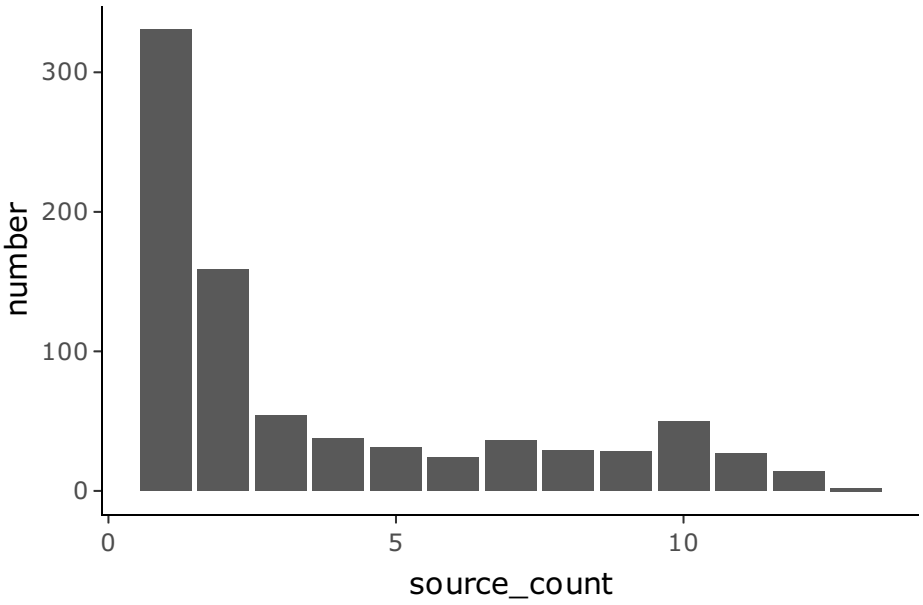
2.2 Bar plot of PanelApp results

Bar plot of the PanelApp analysis.



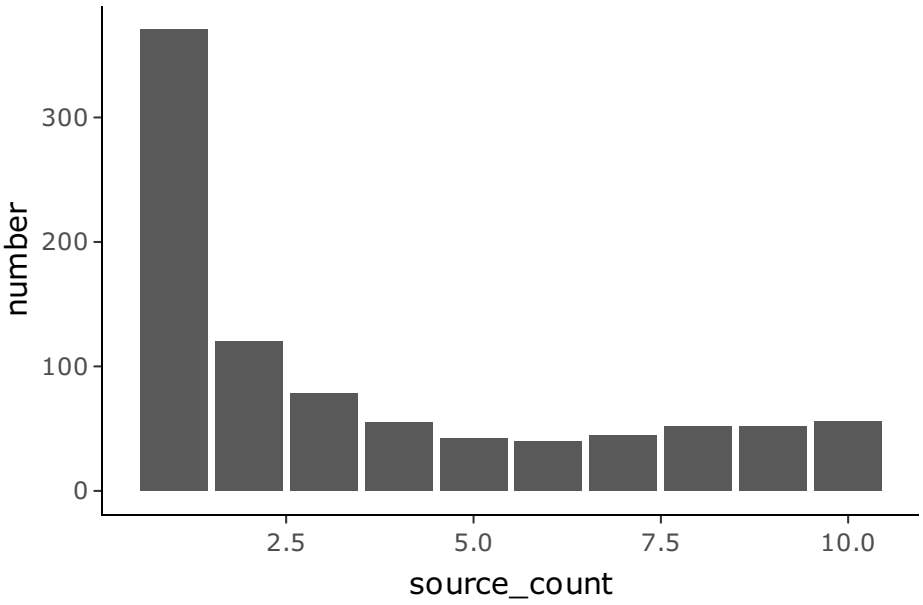
2.3 Bar plot of Literature results

Bar plot of the Literature analysis.



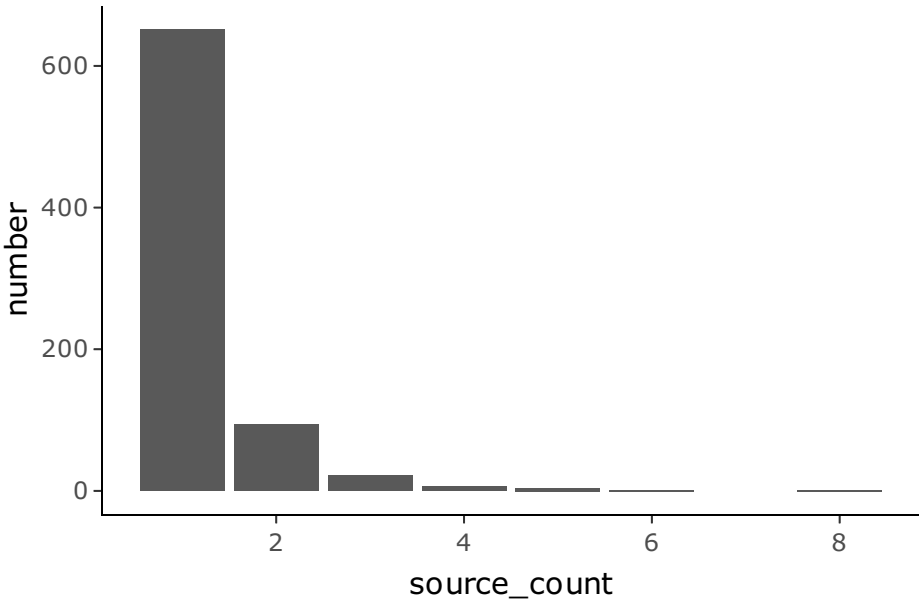
2.4 Bar plot of Diagnostic panels results

Bar plot of the Diagnostic panels analysis.



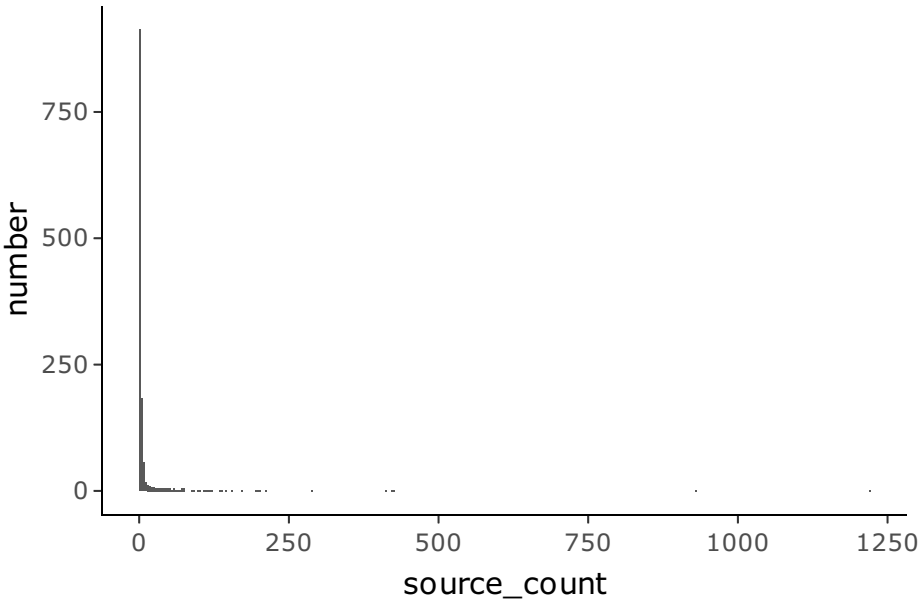
2.5 Bar plot of HPO in rare disease databases results

Bar plot of the HPO in rare disease databases analysis.



2.6 Bar plot of PubTator results

Bar plot of thePubTator analysis.


number
750
500
250
0
0
250
500
750
1000
1250
source_count