A machine-compiled database of genome-wide association studies

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Tens of thousands of genotype-phenotype associations have been discovered to date, yet not all of them are easily accessible to scientists. Here, we describe GWASkb, a machine-compiled knowledge base of genetic associations collected from the scientific literature using automated information extraction algorithms. Our information extraction system helps curators by automatically collecting over 6,000 associations from open-access publications with an estimated recall of 60–80% and with an estimated precision of 78–94% (measured relative to existing manually curated knowledge bases). This system represents a fully automated GWAS curation effort and is made possible by a paradigm for constructing machine learning systems called data programming. Our work represents a step towards making the curation of scientific literature more efficient using automated systems.

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Genome-wide association studies (GWAS) are widely used for measuring the effects of genetic variants on human traits. About 2500–3000 studies have been performed to date, their results are used to estimate disease risks, to understand the function of specific genomic regions, and to train algorithms that predict the effects of new variants.

Most applications require the GWAS associations to be accessible in a structured format amenable to automated analysis by a computer. Several manual curation efforts are underway to catalogue published GWAS associations into structured databases; however, these efforts require time, domain expertise, and can be prone to errors. As more studies are published, the cost of curation is expected to increase.

Here, we describe GWASkb, a machine-compiled knowledge base of thousands of genotype–phenotype associations. It represents a fully automated GWAS machine curation effort, made possible by a paradigm for constructing machine learning systems called data programming. GWASkb is constructed from 589 open-access GWAS publications, and recovers >6000 associations from these publications at an estimated recall of 60–80% and with an estimated precision of 78–94% (both depending on stringency criteria and measured relative to existing manually curated knowledge bases over the same input dataset).

GWASkb is useful to curators as it provides a large dataset of candidate associations for inclusion into existing knowledge bases. These associations are also useful to scientists and clinicians in order to study the genetic basis of human traits and to estimate the disease risks of individuals. To facilitate these use cases, we are making available the code used to create GWASkb, and we also provide an online tool for browsing the associations found by the system. More broadly, our work may form the basis for further efforts to curate Mendelian variants and other biological information.

Results
Automating biomedical literature curation with GWASkb. At a high level, we extract genotype–phenotype relations from the biomedical literature and place them in a structured database (Fig. 1). A typical association consists of a genetic variant, its associated phenotype, and a p-value indicating the significance of the association (see Supplementary Note 3). GWASkb collects these three specific characteristics. Associations also possess additional properties that our system does not yet process; these include an effect size, a risk allele, a target population, and others. Finally, we support our findings with evidence from publications (identified by their Pubmed ID), which are locations inside the document.

When reporting phenotypes, human-curated databases can be at times very specific (e.g., high systolic blood pressure) and at other times less so (e.g., heart disease). In GWASkb, we report simple and precise phenotypes; the former is a high-level description that applies to every variant in the paper (e.g., effects of proteins on inflammation), while the latter is a detailed description that, when available, applies to specific variants (e.g., the name of a specific protein).

We also aim to collect a large set of associations that can be refined by users according to their needs. This approach offers more flexibility than collecting only high-confidence relations, a common approach for manual curation efforts.

Creating GWASkb using IE algorithms. We have structured the system used to generate GWASkb into a set of five components that extract three key pieces of information: genetic variants, their phenotypes, and their p values.

The first component of our system parses the title and abstract of every paper to identify a simple phenotype that will be associated with all its variants. The second component parses the body of the paper to find tuples of Reference SNP cluster IDs (RSIDs) and their associated precise phenotypes. Often, the precise phenotype is abbreviated (e.g., body mass index (BMI)) and a third component attempts to resolve these abbreviations (e.g., BMI). A fourth component extracts p values in the form of (rsid, p value) tuples. Finally, the fifth component constructs a single structured database from all these results.

The components of our information extraction (IE) system are composed of three stages: parsing, candidate generation, and classification (Fig. 2). Parsing is performed with Snorkel—a knowledge base construction framework for documents with richly formatted data (data expressed via textual, structural, tabular, and/or visual cues), such as XML documents. Content is first parsed for structure—the XML tree is traversed and converted into a hierarchical data model with text assigned to tables, cells, paragraphs, sentences, etc. Then, each sentence or cell is parsed for content using the Stanford CoreNLP pipeline, which performs sentence tokenization, part-of-speech tagging, and syntactic parsing. In candidate generation, we identify in the text mentions of some target relation (e.g., p value/rsid pairs). Regular expressions or dictionaries are used to identify candidates that may be valid instances of the relation we are looking for (erring on the side of high recall over high precision). Finally, in the classification stage, we determine which of these candidates are actually correct relation mentions using a machine learning classifier. We use a Naive Bayes classifier with a small number of

![Fig. 1](image-url) The automated information extraction system used to compile GWASkb. The GWASkb system takes as input a set of biomedical publications retrieved from PubMed Central (left) and automatically creates a structured database of GWAS associations described in these publications (right). For each association, the system identifies a genetic variant (purple), a high-level phenotype (pertaining to all variants in the publication), a detailed low-level phenotype (specific to individual variants, if available; red), and a p value (orange). Acronyms are also resolved (red)
hand-crafted features (between 4 and 12), and we train the model using the recently proposed data-programming paradigm.9

One of the most significant bottlenecks in developing machine learning-based IE systems is collecting large sets of hand-labeled training data. Data programming is a paradigm for training models using higher-level, less precise supervision to avoid this bottleneck.9 In this approach, users write a set of LFs: black-box functions that label data points, and that can subsume a wide variety of heuristic approaches such as distant supervision12—where an external knowledge base is used to label data points—regular expression patterns, heuristic rules, and more. These LFs are assumed to be better than random, but otherwise may have arbitrary accuracies, may overlap, and may conflict. A generative model is used to learn their accuracies and correlations from unlabeled data. The predictions of this model can then be used for classification, or to generate labels for a second, discriminative model. For further details see Supplementary Note 1.

In this work, we use data programming to train a generative Naive Bayes classifier over a small number (4–12) of hand-crafted LFs (Supplementary Note 6). We then directly apply these probabilistic labels as predictions. We refer the reader to the appendix for more details.

Reproducibility. In order to make our results fully reproducible, we have released Jupyter notebooks that can be used to generate GWASkb and recreate most of our figures and tables. The notebooks and the source code used to generate GWASkb is freely available on GitHub at github.com/kuleshov/gwaskb.

In addition, we have built an interactive website (see Supplementary Note 10) that enables users to browse associations that have been extracted in GWASkb. Users can search the data by study, phenotype or variant rsid. The entire dataset can also be downloaded from GitHub in CSV format or using the link provided in Supplementary Note 11.

Machine reading helps automate GWAS curation. We compiled GWASkb from 589 open-access GWAS papers, which are papers that are not affected by copyright restrictions that limit our right to perform automated text mining. These papers represent approximately 25% of studies recorded at the time of writing in the NHGRI-EBI GWAS Catalog, a popular human-curated database. We retrieved these papers from the PubMed Central (PMC) repository in XML format and passed the XML source code as input to the IE system. Note that our system can also be deployed on non-open-access papers if a user has legal rights to do so.

Genome-wide associations are typically identified in a discovery cohort and then replicated in a separate replication cohort. Some curation projects (such as GWAS Catalog) only include associations that have been successfully replicated, while others (such GWAS Central) tend to include most associations. GWASkb follows the latter approach; this offers more flexibility and allows researchers to refine the data according to the level of confidence that best suits their needs.

For the purpose of evaluating the precision and recall of our system, we formed a dataset of all automatically extracted associations that were determined to be significant at \( p < 10^{-5} \) in at least one experiment in the study (such as in one cohort or one statistical model). This criterion recovered a significant number of associations present in existing databases, while maintaining sufficiently high precision (Table 1).

It is important to note that our inclusion criterion is different from the one used by databases such as the GWAS Catalog, which typically includes associations that are significant in a combined discovery and replication cohort, unless only discovery data are available and no replication was attempted. Our criterion approach of accepting all associations with their metadata is more flexible, as it allows researchers to refine the data according to their needs. A disadvantage of this approach is that it also includes low-confidence associations, such as ones that have not been replicated, that originate from an earlier study, or that may arise from non-GWAS experiments. A lower-confidence dataset may still be useful for certain applications, such as for testing

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**Table 1 Numbers of associations contained in different GWAS databases; statistics are over open-access papers**

| Database         | Papers | Associations | Unique associations |
|------------------|--------|--------------|---------------------|
| GWAS Catalog     | 589    | 8384         | >2026               |
| GWAS Central     | 516    | 5914         | >364                |
| GWASkb (ours)    | 589    | 6231         | >2777               |

_unique associations are contained in one database and in none of the others. Human curated databases (GWAS Catalog and GWAS Central) significantly differ in their scope. Our machine-compiled repository (GWASkb) automatically recovers a large fraction of known results and also finds a comparable number of unique associations.

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Fig. 2 General structure of a GWASkb system module. The system contains separate modules for extracting variants, phenotypes, and values, and for resolving acronyms. Each module consists of three stages. At the parsing stage, we process papers using the Stanford CoreNLP pipeline, performing full syntax parsing. Next, given a target relation (e.g., variant-phenotype), we generate a large set of candidates, some of which could be correct instances of the target object on relation. Then, at the classification stage, we determine which candidates are correct using a machine learning classifier.
whether certain pathways are enriched for associated variants. However, this also requires manually filtering variants that do not meet the significance threshold for other applications, which can be burdensome. To assist with this process, we are releasing metadata that helps identify the cohort associated with a given variant (see Supplementary Note 9). This metadata can later be used to train classifiers that automatically identify the target cohort.

GWASkb recovers up to 80% of manually curated associations. GWAS Central and GWAS Catalog contain, respectively 3008 and 4023 accessible associations linked to the 589 open-access studies. These associations are defined as tuples of PubMed ID, variant RSID, phenotype, and p value for which the RSID is contained in the open-access XML content made available through PubMed Central. For GWAS Catalog we use the reported trait for our analysis rather than the ontology terms (EFO). To measure recall relative to human-curated databases, we need to determine whether each (PubMed ID, RSID, phenotype, p value) tuple reported in GWASkb is also present in the human-curated database. This requires deciding whether phenotypes reported in GWASkb are equivalent to ones reported by human curators; to determine this, we manually created a mapping between GWASkb phenotypes and phenotypes reported in either GWAS Central or GWAS Catalog for the same PubMed ID and RSID (see Methods).

Since databases use different levels of precision to describe traits (e.g., smoking behaviors vs. cigarette packs per day), we also specify whether our reported phenotype is exact or approximate; in the latter case, it is still useful, but lacks some detail. Table 2 contains examples of relations contained in GWASkb at different levels of precision.

The dataset reported in GWASkb contained 2487 (82%) associations from GWAS Central with approximately correct phenotypes, as well as 3245 (81%) associations from the GWAS Catalog. It also recovered 1890 (63%) associations from GWAS Central and 2762 (69%) associations from GWAS Catalog with full accuracy on the phenotype. Some associations were not correctly recovered because the GWASkb reported phenotype was incorrect: 89 (3%) for GWAS Central and 147 (4%) for GWAS Catalog. In the remaining cases, we were not able to report the variant itself. The main causes of this are when the variants are expressed only in the text and not in tables, or when the format of the table is particularly difficult to parse (e.g., when multiple RSIDs and p values are reported in the same row). Overall, GWASkb recovered 81–82% of manually curated associations at a level of quality that will be useful in many applications.

Machine curation uncovers useful new associations. In total, GWASkb contains 6422 associations within the 589 input papers, 2959 (46%) of which could not be mapped to GWAS Catalog or GWAS Central. We investigated this further by first manually inspecting a random subset of 100 novel associations (with independent validation from two independent annotators). We found that 88 associations fully met the specifications of our system, 7 were incorrect, and 5 were originally identified by a different study (and referenced as background material). Most of the errors of our system can be attributed to incorrect phenotypes.

Of the 88 associations matching system specifications, 44 were not significant at $10^{-5}$ in all cohorts, hence were excluded from GWAS Catalog for scientific reasons. We report these variants because they may still be useful in applications in which a noisy dataset is acceptable. Another 36 were excluded because they were in the same locus as a more significant variant; however, these were generally not in perfect linkage disequilibrium (LD) and 27 were in weak LD with the GWAS Catalog variant ($r^2 < 0.5$ as determined by the LDLink tool). We argue for cataloguing these variants, as the LD cutoff for what constitutes a significant variant may depend on the scientific application. Another eight variants were not included because they were determined to be significant in both the target study and an earlier study (note that GWAS Catalog guidelines state that such variants should be catalogued).

LD between new and existing variants. To validate the novel variants found by our system, we conducted a series of analyses aimed at characterizing the variants’ function.

First, we reasoned that detected variants may be in LD with known variants (because they originate from the same LD block), or among themselves, thereby inflating our number of truly novel associations. We estimated LD from the Thousand Genomes dataset (see Supplementary Note 2); Fig. 3 shows the histogram of $r^2$ distances between each novel variant, and its closest variant in the GWAS Catalog. The distribution of $r^2$ scores is highly multimodal, with large peaks at $r^2 = 1$, and many more at $r^2 = 0$.

Using a threshold of $r^2 > 0.5$, we filtered our set of new [pmid, rsid, phen, pvalue] associations from 3170 to 1494 by removing variants in LD with known manually curated variants; of the 1676 variants that we eliminated, 765 were not in the 1000 Genomes database or their closest previously known variant was not in the database; the remaining 911 single-nucleotide polymorphisms

### Table 2 Examples of associations identified by GWASkb

| Study | Association | Simple phenotype (GWASkb) | Precise phenotype (GWASkb) | p Value (GWASkb) | Phenotype (GWAS Cat) | p Value (GWAS Cat) |
|-------|-------------|----------------------------|----------------------------|-----------------|----------------------|-------------------|
| Genome-wide pharmacogenomic study of metabolic side effects to antipsychotic drugs | rs17661538 | Antipsychotic drugs/metabolic side effects | Clozapine—Triglycerides | 1.00E−06 | Clozapine-induced change in triglycerides | 1.00E−06 |
| Genome-wide meta-analysis identifies seven loci associated with platelet aggregation in response to agonists | rs2566888 | Platelet aggregation | - | 5.00E−19 | Platelet aggregation, and epinephrine | 5.00E−19 |
| A genome-wide association study of the Protein C anticoagulant pathway | rs13730255 | Protein C | funcPS | 3.00E−06 | Anticoagulant levels (funcPS) | 3.00E−06 |

Associations can be classified as correct (rs17661538), partially correct (rs2566888; the precise phenotype is missing) and incorrect (rs13730255). We also compare these associations to their corresponding entries in the GWAS Catalog.
SNPs were in LD with known variants. We further reduced this set to 1304 associations by eliminating novel variants that were in LD with each other. Thus, although many variants are in LD with known variants, over 40% of our discovered variants do not appear to be linked to variants previously identified in GWAS databases.

Although our system reported multiple variants from the same LD block, these variants may still be useful, since we do not know which variant an LD block is truly causal and the $r^2$ cutoff for defining LD blocks is somewhat arbitrary and may vary. We believe that filtering should be performed by the user, depending on their goal; this is also the approach taken by the GWAS Central repository.

Comparison to other approaches for estimating significance. Our second analysis focuses on the biological function of the novel variants. We focus on two large classes of phenotypes: neurodegenerative diseases (ND; 27 traits, including Autism, Alzheimer’s, Parkinson’s, etc.) and autoimmune disorders (AI; 23 traits, including Diabetes, Arthritis, Lupus, etc.); for the analyses below, we consider the subset of variants that are not in LD with any variant in the GWAS Catalog or GWAS Central (283 ND SNPs and 155 AI SNPs).

We also collected two sets of genes that were found to be highly expressed in brain cells as well as in blood cells; specifically, we reasoned that SNPs associated to neuropsychiatric and autoimmune diseases should be more highly enriched near genes expressed in brain and immune cells, respectively. Indeed, we found that variants associated with ND diseases (32 ND SNPs in total) occurred significantly more often within 200 kb of genes with preferential brain expression, while variants associated with AU traits (15 variants in total) were found more frequently near genes with preferential blood expression ($\chi^2$ test: $p < 0.05$; see Supplementary Note 2).

We should note, however, that the vast majority of ND and AU variants were found far from coding regions. To test whether this set of SNPs also make biological sense, we used GREAT$^{13}$, a tool which annotates the function of variants in intergenic areas of the genome. In particular, GREAT links intergenic regions with Disease Ontology (DO) terms, and outputs terms that are significantly enriched for a particular set of variants. When we applied GREAT to ND SNPs, we found a strong enrichment in regions known to play a role in ND-related phenotypes, such as cognitive disease ($p < 10^{-32}$), dementia ($p < 10^{-23}$), and neurodegenerative disease ($p < 10^{-23}$). Similarly, AI variants were significantly associated with AI-related terms, the most significant of which were disease by infectious agent ($p < 10^{-27}$), viral infectious disease ($p < 10^{-19}$), and autoimmune disease ($p < 10^{-17}$). In fact, the top 20 DO terms for each ND traits were all exclusively associated with the correct family of phenotypes (Supplementary Tables 1 and 2). Hence, our predicted variants were consistent with external annotations.

Examining the effect sizes of novel GWASkb variants. Finally, we analyzed the magnitude with which novel variants affect their predicted phenotypes and other, related traits. Specifically, we used freely available GWAS summary statistics from the LD Hub project$^{14}$ to assess the distribution of SNP effect sizes across novel variants and compared them to those of random SNPs. We focused on the 11 most frequent traits in our dataset for which summary statistics were available; for each trait, we identified an LD Hub study that provides effect sizes (in the form of beta coefficients or log odds ratios) for that trait. Figure 4 compares the distribution of effect sizes of the novel variants identified in GWASkb to the distribution of effects sizes for all SNPs, again restricting to variants that show no LD with other variants in GWAS databases. Whereas the distribution of random SNPs is centered around zero, as one would expect, novel SNP effect sizes appear to follow a different distribution (Kolmogorov–Smirnov test; see Fig. 4 and Supplementary Figs. 1 and 2) and tend to have significantly higher magnitudes than expected.

Discussion
Curation of the literature is critical because if GWAS associations are not recorded in a database, they are effectively missing for many practical purposes, such as for training machine learning systems to predict SNP function. GWAS studies are also costly (often involving genotyping tens of thousands of subjects), and it thus a waste of research funding to not fully record their results. Systems like the one used to create GWASkb can assist the curation process by providing useful candidates to human curators.

Most existing GWAS databases are constructed by human curators, who are expert scientists with advanced training that enables them to understand and parse complex study designs. Manual curation yields accurate and trusted results, albeit at a high labor cost (e.g., GWAS Catalog associations are verified by a second curator for maximum accuracy). An alternative to curation is to ask authors to directly report their findings online. This is already possible within GWAS Central, although in practice few authors choose to do this. In addition, past studies still need to be curated. An ideal solution appears to involve a combination of authors, machines, and curators.

However, manual-curation is a difficult task, and can miss certain associations. Curating papers is often a tedious task involving browsing through highly technical material in search of
short snippets of text. Humans are generally not well-suited to this kind of work: they may accidentally skip table rows, or become tired and skip a paragraph. Curation also requires understanding advanced technical concepts such as LD or multiple hypothesis testing. This makes the task unsuitable for crowdsourcing approaches.

Computers, on the other hand, do not suffer from the aforementioned limitations: they excel at repetitive work and only need to be programmed by experts once. Crucially, even though machines make errors, these errors are systematic, not random: one may follow an iterative process of fixing these errors and redeploying the system, until a sufficient level of accuracy is reached. Redeploying our system takes on the order of hours, while asking humans to return and correct their errors would take at least months.

Of course, humans also have many advantages over machines. Indeed, the sets of GWASkb and human-curated associations were quite distinct. The most accurate and complete GWAS database is in fact a combination of both sources. In the future, our system can be further improved by extracting additional information about variants (e.g., risk alleles and odds ratios). In addition, the current version of our database does not contain crucial study metadata such as study design, study stage, ancestry information, statistical methodology, etc. These are typically curated by human experts.

In summary, we have introduced a machine reading system for extracting structured databases from publications describing genome-wide association studies. Our results represent a step towards using machine reading algorithms to help human curators synthesize knowledge in the biomedical literature, helping make GWAS research faster and more accurate.
**Methods**

**Detailed description of the GWASkb system.** The system used to create GWASkb is implemented in Python on top of the Snorkel IE framework. Snorkel provides utilities for parsing XML documents and training machine learning classifiers. The GWASkb system extends the parsers/classifiers in Snorkel and applies them to the GWAS extraction task. Below, we provide additional details on the various components of the system.

To identify simple phenotypes, we start by parsing paper titles and abstracts and generate candidates from the EFO, Snomed, and Mesh ontologies. We use 11 labeling functions (LFs), which include the following: is the mention in the title; is the mention less than five characters; does the mention contain nouns; is the mention in the first half of the sentence, etc. We include the full list of LFs in Supplementary Note 6. The high-level phenotype is the set of three highest scoring mentions exceeding a user-specified score threshold or the single highest mention if none exceeds the threshold; this enables us to handle multiple valid phenotypes.

To identify precise phenotypes, we start by only parsing tables and generate candidates from cells whose header contains the words “phenotype”, “trait”, or “outcome”. Candidate p values are generated by matching a regular expression; candidate relations consist of horizontally aligned phenotype and p value candidates. We use three LFs (provided in Supplementary Note 6): is the candidate mostly a number; is the header of the cell (indicating it is in a phenotype column) very long; does the mention contain words referring to an rsid.

Next, we resolve acronyms by looking at the entire paper, including tables and the main natural language text in the body of the paper. We extract candidates from aligned pairs table cells, where one row is labeled “phenotype”, “trait”, or “description”, while the other is labeled “abbreviation”, “acronym”, or “phenotype”. We generate candidates from the main text using a regular expression. Our LFs, include the following: is the candidate all in uppercase characters; does the candidate consist of the letters of each word in the Snomed dictionary; does the acronym candidate consist of the letters of each word of the phenotype candidate; is one a prefix of the other; etc. The module for resolving abbreviations is linked in Supplementary Note 7.

Finally, we identify p values by again generating candidates from tables; SNP candidates are generated using a regular expression; p value candidates are ones that match one of three regular expressions (see Supplementary Note 8); candidate relations consist of horizontally aligned SNP and p value candidates (with at most one rsid per row). These candidates were accurate and we report them all.

**Mapping phenotypes across databases.** In order to compare against GWAS Central and GWAS Catalog, we define mappings between GWASkb phenotypes and ones used in these repositories. These mappings are tables with about 800 entries each that also indicate whether the mapping is fully or partially correct (e.g., “smoking behaviors” is less precise than “packs per day”). We define the latter as conceptually containing the precise label while also being not so broad as to be useless. See also our earlier discussion on high- and low-level phenotypes.

**Understanding the errors of GWASkb system components.** Errors at the simple phenotype extraction stage mostly occur when the true phenotypes are not found in our candidate dictionaries (e.g., for the phenotype “genome-wide association study in bipolar patients,” we can only generate the candidate “bipolar disorder”). The second major source of error are phenotypes mentioned only in passing (e.g., “proteinase inhibitor 5 precursor”), and they are presented in tables with confusing symbol is not clearly related to the full expression (e.g., CYS5 for Cysteine proteinase inhibitor 5 precursor). Errors at the simple phenotype extraction stage mostly occur when the true phenotypes are not found in our candidate dictionaries (e.g., for the phenotype “phenotype whose association is being reported”). Then, we look at the fraction of these relations whose phenotype is useless. See also our earlier discussion on high- and low-level phenotypes.

To resolve acronyms, we consult the entire paper, including tables and the main natural language text in the body of the paper. We extract candidates from aligned pairs table cells, where one row is labeled “phenotype”, “trait”, or “description”, while the other is labeled “abbreviation”, “acronym”, or “phenotype”. We generate candidates from the main text using a regular expression. Our LFs, include the following: is the candidate all in uppercase characters; does the candidate consist of the letters of each word in the Snomed dictionary; does the acronym candidate consist of the letters of each word of the phenotype candidate; is one a prefix of the other; etc. The module for resolving abbreviations is linked in Supplementary Note 7.

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**Estimating the precision of GWASkb**

We estimate our overall precision at 94% relative to the output specifications of our system. Of the 6422, associations reported by our system, we consider 3463 to be correct because we could confirm them in an existing database (GWAS Central or GWAS Catalog). We estimate the error rate on the other 2999 relations to be between 12% (incorrect and repeat relations; this corresponds to GWASkb specifications) and 53% (when adding the 44 variants not confirmed in the meta-analysis but included in at least one of the other repositories to our set of errors). For an estimated total precision of 78–94% over the 6422 reported relations.

**Data availability**

The complete datasets and code used in the current study are available in the gwaskb repository, accessible at [https://github.com/kuleshov/gwaskb](https://github.com/kuleshov/gwaskb). The resulting knowledge base, GWASkb, is also accessible via a web portal at [http://gwaskb.stanford.edu/](http://gwaskb.stanford.edu/). All other data are contained within the article and its supplementary information (the source data folder contains source code, raw input data including papers and ontologies, extra figures, extra Jupyter analysis notebooks; see Supplementary Note 5).

**Code availability**

The complete datasets and code used in the current study are available in the gwaskb repository, accessible at [https://github.com/kuleshov/gwaskb](https://github.com/kuleshov/gwaskb), which includes full documentation for running the code and reproducibility data.
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Author contributions

V.K. conceived the study. B.H., A.R., and V.K. developed the modules for the Snorkel system. V.K. developed the GWASkb system. B.H., J.D., and C.V. performed the computational analysis. J.D. developed the web interface. V.K., B.H., A.R., and Y.L. wrote the paper. Y.L., C.R., S.B., and M.S. supervised the study.

Additional information

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