



Recommendations of the DNA Commission of the International Society for Forensic Genetics (ISFG) on quality control of autosomal Short Tandem Repeat allele frequency databasing (STRidER)



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ABSTRACT

The statistical evaluation of autosomal Short Tandem Repeat (STR) genotypes is based on allele frequencies. These are empirically determined from sets of randomly selected human samples, compiled into STR databases that have been established in the course of population genetic studies. There is currently no agreed procedure of performing quality control of STR allele frequency databases, and the reliability and accuracy of the data are largely based on the responsibility of the individual contributing research groups. It has been demonstrated with databases of haploid markers (EMPOP for mitochondrial mtDNA, and YHRD for Y-chromosomal loci) that centralized quality control and data curation is essential to minimize error. The concepts employed for quality control involve software-aided likelihood-of-genotype, phylogenetic, and population genetic checks that allow the researchers to compare novel data to established datasets and, thus, maintain the high quality required in forensic genetics.

Here, we present STRidER (<http://strider.online>), a publicly available, centrally curated online allele frequency database and quality control platform for autosomal STRs. STRidER expands on the previously established ENFSI DNA WG STRBASE and applies standard concepts established for haploid and autosomal markers as well as novel tools to reduce error and increase the quality of autosomal STR data. The platform constitutes a significant improvement and innovation for the scientific community, offering autosomal STR data quality control and reliable STR genotype estimates.

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1. Status quo of autosomal STR quality control and databasing

Short Tandem Repeats (STRs), also known as microsatellites, are polymorphic DNA regions that are widespread throughout the human genome [1]. They typically consist of simple, compound or complex DNA motifs that are 2–7 base pairs (bp) in length and

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show high variability between unrelated individuals. The forensic field adopted autosomal STRs in the early 1990s [1], and STRs have become the primary workhorse for individual identification in criminal casework, kinship analyses, and identification of missing persons [1–3]. Forensic STR loci were selected according to stringent criteria, and sets of core loci were defined. These loci largely overlap between countries [2].

Forensic evaluation of STR-based genetic evidence requires (i) correctly called and reported STR alleles and genotypes, and (ii) unbiased estimates of STR allele frequencies supported by high quality raw data and exclusion of close relatives among donors. Numerous guidance papers on forensic STR typing and interpretation were published (e.g., [4–10]) that highlight the importance of internal and external quality control (QC). Community based (e.g., [11–14]) and commercial (see example in [15]) proficiency testing programs that are powerful in highlighting problems with the analytical procedure and interpretational issues of STR typing in a routine forensic laboratory have been implemented. However, published datasets that form the basis for statistical evaluation in forensic genetics are not yet part of any external standardized quality control program – despite a growing number of contributions. Published datasets may be fraught with error and, thus, not meet forensic requirements.

In the fields of haploid markers, mitochondrial DNA (mtDNA) and Y-chromosomal STRs, the need for centralized independent QC was quickly recognized. QC tools were put in place in conjunction with databasing efforts (EMPOP at <http://empop.online> [16] and YHRD at <http://yhrd.org> [17]) to avoid the publication of erroneous data [16,18,19]. The editors of the leading forensic genetic journals; *Forensic Science International: Genetics* [20] and *International Journal of Legal Medicine* [21] require QC of mtDNA and Y-STR population data prior to submission of the research papers to the journals. This QC process involves plausibility (i.e. the likelihood of the reported genotypes under a relevant hypothesis, e.g. a given population, etc.) and phylogenetic checks with the aid of customized software that highlights data ambiguities and potential errors. The curators of the QC programs initiate dialogues with the authors of the submitted datasets to discuss the observations and produce a final version of the dataset that is then submitted to the journal alongside the manuscript. Successful QC is documented by an accession number that serves as the unique identifier for the dataset and confirms completion of the data review. These strategies have significantly improved overall data quality in forensic genetics and reduced the amount of errors that would have otherwise been mistakenly introduced in scientific publications (W. Parson and L. Roewer, personal communication).

Such QC strategies are lacking for the field of autosomal forensic STR studies, in part because phylogenetic studies are more difficult for diploid markers. Another reason may be the lack of a centralized STR database that can provide QC tools based on a set of qualified STR data. Among the publicly available forensic STR allele frequency databasing initiatives, the following are briefly presented:

- (i) The European Network of Forensic Science Institutes (ENFSI) DNA working group STR population database (STRbase; <http://strbase.org>) can be used to calculate STR profile match probabilities in European populations by single or batch queries. Adjustment factors are offered. In its current second version, STRbase holds up to 5,179 high quality profiles of 16 STR loci [6] from 19 countries [8]. Allele frequencies per marker and population are offered for download, while the STR profiles cannot be accessed. The data have been subject to rigorous QC as documented in [22].
- (ii) The National Institute of Standards and Technology (NIST) STR DNA Internet DataBase (STRBase; <http://www.cstl.nist.gov/>

[strbase](http://strbase.org)) acts as a forensic information resource that provides details on observed common and rare alleles, annotated sequences, analytical technologies (including commercial kits, primer sequences, and validation studies), lists involved organizations and scientists, and consolidates and organizes STR literature. Allele frequency tables or profile frequency estimates cannot be generated, but the OmniPop program is linked for download to calculate the latter metrics using data published prior to 2004 from >200 populations and 9–16 loci. The results are also displayed on a world map [23].

- (iii) ALFRED, the ALlele FREquency Database (<https://alfred.med.yale.edu/>), is a comprehensive database of allele frequencies (and related information) compiled from >660,000 individuals, including all core forensic STRs. Although data is held for over 700 populations, the number of forensic STR variation studies varies from six populations for D1S1656 to 432 for TH01. Nevertheless, ALFRED provides extensive allele frequency data for a large number of populations outside Europe in the majority of commonly used forensic STRs. Frequencies can be displayed in stacked-bar format, on maps, and can be downloaded [24].
- (iv) *pop.STR* (<http://spsmart.cesga.es/popstr.php>) is an STR allele frequency browser including data from 70 STR loci and 57 populations (comprising 3,809 samples in total). Various populations or population groups can be combined in up to five user-defined sets per query. From these queries, *pop.STR* presents allele frequencies in bar charts and summarizes the variation data into common population genetics indices. Summary allele frequency data, but not genotypes, can be downloaded in CSV file format for data processing by the user [25].
- (v) PopAffiliator (<http://cracs.fc.up.pt/~nf/popaffiliator2>) is an online calculator for individual affiliation of an STR genotype to five major population groups based on >56,000 profiles [26].
- (vi) ALLST*R (<http://allstr.de>) is a commercial website collecting published and unpublished allele frequency data from >200 populations and >90 STR markers. It provides genetic indices, information on loci, populations, mutation rates, and literature references. Datasets can be added and exported and receive a unique identifier.

None of these databases provides feedback on data quality or enables QC of STR datasets as outlined for haploid markers above. This limitation was already addressed in 2008 along with another STR database effort at the time that relied “on the acceptance criteria used by the main forensic international journals” [27]. Therefore, we suggest implementing a locally curated, freely-accessible STR database following the successful models of EMPOP and YHRD to provide a centralized archive of data serving as platform for the provision of probability estimates, specific software, and for the development of novel tools for data QC and other forensically relevant analyses.

2. STRidER: a novel STR database providing QC software tools

2.1. Rationale and concept

Available STR population datasets potentially contain errors that were not addressed or happened during the publication process. Some of such errors are easy to spot, such as incorrect allele nomenclature (e.g., “22.4” in a tetrameric STR marker) or incorrectly prepared frequency estimates (e.g., where the reported STR allele frequencies for a marker do not add up to 1) (see [28]). In just one example, re-typing of a widely applied population dataset after 16 years revealed a certain number of clerical, technical, and

data/sample processing errors [29]. Undoubtedly, reviewers and editors cannot easily identify all possible inaccuracies in a table of allele frequencies in a submitted manuscript, neither can nor should the readers do so in published papers. Experiences in the field of haploid markers have shown that outsourced expert QC using appropriate software tools takes the responsibility of *ad hoc* quality checks, aids the process of data review and results in high quality published data [16,17]. The field of mtDNA analysis was at the center of a high-profile dispute about the quality of the published data in the mid-2000s (see [30–34]) and has strongly benefited from the centralized QC program associated with EMPOP [16,21,35,36]. In this manuscript, we suggest that the established scrutiny concept can be adopted for autosomal STR data. We present STRidER (STRs for Identity ENFSI Reference Database), an expanded and enhanced version of the ENFSI STRbase, which continues to serve as a high quality STR population database enabling scientifically reliable frequency estimates. STRidER takes the innovative step of providing QC of autosomal STR data in a standardized fashion by taking advantage of established and newly-developed tools. Datasets will be subjected to a number of plausibility checks to prevent many of the errors currently encountered and thus ensure more reliable allele frequency estimates. Direct communication between STRidER and authors of population data will facilitate and accelerate corrections as well as lead to an improvement of the overall quality of published datasets. The reviewers will be provided with a quality-checked STR dataset indicated by accession numbers, allowing the reviewers to concentrate on the manuscript content beyond the genotype data itself such as the relevance and composition of the sample set given the available information on the population(s) under study. Finally, the accepted data will become rapidly available in a centralized online database of autosomal STRs used in forensic genetics, instead of being dispersed in numerous journal publications.

2.2. Data analysis and QC workflow

2.2.1. Data submission

STRidER is accepting data from diverse worldwide populations and forensically relevant autosomal STR markers for QC, with datasets that comply with ethical standards and the minimum requirements of journals (e.g., 15 STR loci per sample and a minimum sample size of 500 for *Forensic Science International: Genetics* [36]) as well as other data that are not produced for peer-reviewed publication. Population datasets consisting of STR genotypes can be submitted to STRidER by registered users in a standardized form. Example data tables and details on the standardized format are provided via the website (<http://strider.online>). Once accepted and uploaded onto STRidER the complete genotypic profiles will be disassembled. For each locus, the STR allelic pair will be kept intact, while single locus genotypes will be shuffled between donors within the population. This keeps the intra-locus genotype configuration unaltered for QC, but disrupts the inter-locus genotype association in order to protect the privacy of sample donors (see Fig. 1). The original datasets will not be stored in the database. Raw data must be kept in the contributing research group and might be requested for inspection.

For data handling and QC statistics, information beyond the genotypes has to be provided at submission (included in the downloadable data submission template):

- General information
 - Submitting institution/laboratory, contact person(s), e-mail address(es)
 - Laboratory accreditation status

Sample ID	TH01	TH01	vWA	vWA	FGA	FGA
Anonymous 0001	6	9.3	16	17	20	24
Anonymous 0002	7	7	13	14	18	18
Anonymous 0003	8	9	12	16	22	24
Anonymous 0004	9.3	9.3	11	13	19	20

Fig. 1. The principle of shuffling of genotypes in STRidER demonstrated for four samples and three STR loci. For each population, the submitted data are arranged into a table format with rows per individual profile and columns per locus. For each locus, the STR allelic pair is kept intact, but single locus genotypes are shuffled between individuals. This keeps the intra-locus genotype configuration intact, but disrupts the inter-locus genotype association in order to protect the privacy of sample donors. Shades indicate the original genotypes; this information is not stored in STRidER.

- Formal statement on informed consent and institutional review board approval (if applicable)
- Manuscript and tentative journal (if applicable)
- Information on the sample set
 - Type of sample set (e.g., random, “convenience”, casework) and exclusion/inclusion criteria (if any have been used), pinpointing potential bias [37]
 - Type of specimen (blood, buccal swab, other; fresh tissue, FTA cards, etc.)
 - Number of individuals (female/male)
 - Unique anonymized identifier for every sample, not traceable by third parties, for communication during QC (will be deleted afterwards)
 - Country/region/city of donors’ geographic origin in as much detail as possible
 - Donor population and metapopulation [17], e.g., ethnic, tribal or language group(s) and subgroup(s) in as much detail as possible
 - Report of published genetic data from overlapping population samples; concordance (if applicable)
- Information on laboratory analyses
 - DNA extraction method/direct amplification
 - STR typing kit(s) (kit version listed)/homemade multiplexes (allelic ladders utilized)
 - Detection platform used and settings applied
 - Detection chemistry, e.g., polymer type
- Information on data analysis and handling
 - Analysis software (software version listed), settings, peak detection thresholds, etc.
 - Type of positive control(s) used and pass/fail information (raw data may be requested for QC)
 - Type of data transfer: automated or manual
 - Information on suspected null-alleles and observed discordances if overlapping population samples were typed using different chemistries

2.2.2. General and plausibility checks on submitted STR datasets

The basic QC measures and descriptive plausibility tests listed below may appear trivial. However, experience from haploid markers (see [35]) has indicated their importance: observations are significant for overall data quality and signpost potential idiosyncrasies, errors, technical problems, sampling bias, or population substructure, thus the need for special attention. However, there are no general cutoff values, as genetic properties reflect the history of the population under study, and this must be taken into account.

- Matching and accuracy of information
 - Number of individuals indicated = number of individual genotypes reported

- STR loci included in indicated kit(s) = STR loci in submitted genotypes
- Check on STR locus names
- Completeness of genotype data
 - Two alleles per locus in ascending order; double specification of homozygous alleles
 - F-designation to indicate ambiguous calls, such as *e.g.* 13/F, which indicates that allele 13 has been clearly identified in the raw data and an additional allele may be present that cannot be defined (*e.g.*, due to masking by an artifact, unusual peak shape, very low peak height, etc.); F-alleles will not be used for allele frequency estimates
 - Tri-allelic genotypes are accepted but will not be used for allele frequency estimates
 - Partial profiles will be excluded, since data from low-quality samples is not useful for forensic frequency estimation (as recommended for mtDNA haplotypes [38])
- Plausibility of reported alleles and genotypes
 - Correct allele nomenclature
 - Proportion of not previously observed and rare alleles (below frequencies of 0.01 [39] and $5/2n$ [40] in the population sample)
 - Proportion of off-ladder (compared to the applied allelic ladder), F-, and suspected null-alleles
 - Proportion of matching alleles in pairwise comparisons of complete genotypes – identification of duplicates, duplicates with transcription errors, and close relatives (parents and siblings) [37]
 - Proportion of tri-allelic genotypes
 - Proportion of observed homozygotes
 - Proportion of (unexpected) homozygotes for rare alleles

2.2.3. Further statistical analyses of STR datasets

Additional forensic efficiency and population genetic parameters commonly reported in announcements of STR population data are calculated per locus and/or cumulatively over all loci including expected homozygosity (h), expected heterozygosity (H_E , H_E), observed heterozygosity (H_O , H_O , H), power (probability) of discrimination (PD), power (probability) of exclusion (PE), typical paternity index (TPI), and probability of identity (P_i ; *i.e.* matching or random match probability, PM). In addition, more complex analyses assess the excess of (possibly false) homozygotes, calculate population-specific F_{ST} (θ) values (discussed in [28]), and apply statistical tests for Hardy-Weinberg equilibrium (HWE) conformity to measure independence of alleles at each locus. As outlined, these indices depend on the population's history; therefore, the effects of substructure, inbreeding, selection, biased sampling, analytical problems, errors etc., cannot be distinguished without further investigation. Furthermore, the incidence of inflated homozygosity due to primer binding site mutations causing allele drop-out is commonly population specific, making HWE conformity a critical test of novel population data. Even if neither departure from HWE nor high intra-population F_{ST} are anticipated in a properly made sample from most human populations (except for small, isolated groups), there are no strict "expected" ranges for QC purposes. All analyses are carried out on shuffled datasets and thus limited to intra-locus tests.

Once limits for metric deviations are established, STRidER will outline how to proceed in such cases. Until then, these parameters are generated from the STR data after basic QC for illustrative and exploratory purposes, but can also indicate technical problems in some cases. Further parameters may be added as required.

2.3. STRidER as high quality STR allele frequency database

Validated novel data will be uploaded to STRidER upon journal publication or on individual agreement. At a later stage, previously published datasets will be considered on request or our own initiative and thus contribute to STRidER's growth if they pass (possibly modified) QC. Users will be able to download population-specific allele frequency tables calculated from these datasets, *e.g.*, to feed into other software. Individual STR profiles are neither accessible nor stored in the database; therefore, privacy issues are not violated. The allele frequencies are employed in database queries to estimate population-specific and overall frequencies (random match probabilities) of STR profiles. A minimum allele frequency of $5/2n$ [40] or a fixed value, such as 0.01 [39], can be applied for calculations according to laboratory practice. Correction factors adjusting for minimum allele frequency, potentially undetected population substructure, sampling bias, and relatedness are offered, including Balding's size bias correction [41], Balding and Nichols' F_{ST} correction [42], and the upper bound of a 95% confidence interval [40]. Methods are reviewed in [43]. For more reliable frequency estimates, datasets from the same population will be combined when they present no statistically significant differentiation (from pairwise F_{ST} calculations), the pooled sample conforms to HWE expectations, and non-genetic information such as sampling criteria indicate that they represent the same non-heterogeneous population. Otherwise, samples will be presented separately. Additional allele frequency correction factors and differentiation tests may be added at a later stage.

Accepting allele counts or frequency data instead of individual genotypes would allow STRidER to quickly grow as data repository that could also accept STR mixture information, but would in turn impair some of the QC measures described above intended for a high quality population database.

3. Benefits to the scientific community from services offered by STRidER

The STRidER database and QC platform constitutes a significant augmentation of services accessible to both scientists and practitioners in forensics and beyond. Providing detailed information on high-quality samples, STRidER may serve other fields interested in human genome variation and its geographic patterns, such as anthropology and population genetics. STRidER is offered at no cost to promote data quality, but should be referenced in any publication of validated data. STRidER does not interfere with existing resources described above. To summarize, the STR community will benefit from STRidER in three inter-related ways (illustrated in Fig. 2):

- (1) *Centralized quality control: only high quality autosomal STR data are published*

STRidER aids authors, reviewers, and editors with QC data, providing the possibility to rectify data errors before publication, and thus to ensure a high quality checking procedure that users can trust. Journals may decide to accept autosomal STR data only after QC by STRidER in an analogous approach to that adopted for haploid markers [20,21,36].

- (2) *Databasing: a curated autosomal STR allele frequency database that can be queried*

The STRidER database will continuously grow in numbers of markers and populations only by high quality data contributed by the forensic community. In this way, it will be useful for all forensic

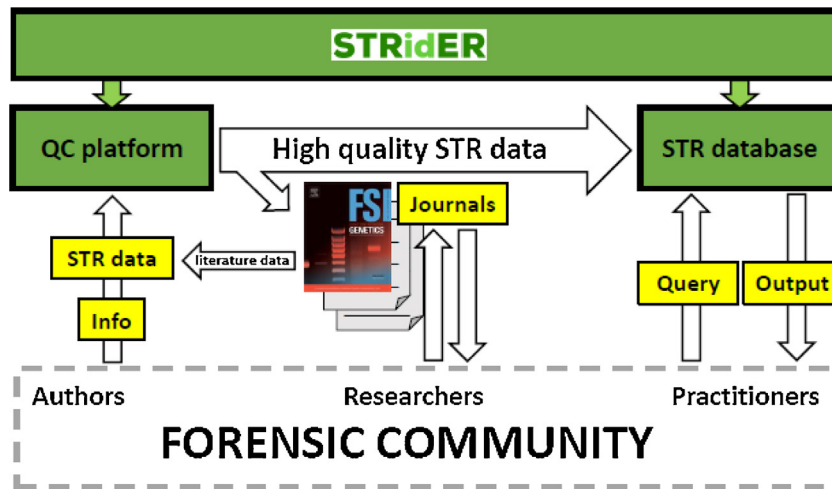


Fig. 2. STRidER in the field of forensic STR typing. STRidER functions as database enabling STR profile frequency estimates and as QC platform for new STR datasets to be checked. Previous experience with haploid DNA markers (mtDNA – EMPOP and Y-chromosomal STRs – YHRD) demonstrates that independent external quality control based on a centrally curated database offers valuable feedback to authors, higher quality datasets for publication, and a reliable basis for frequency estimates.

practitioners for reliable STR profile frequency estimates and the retrieval of allele frequency data. Queries can be filtered for all information associated with the population datasets (as kept in EMPOP) [16].

(3) Transparency and traceability of autosomal STR data: setting standards

Allele frequencies and additional statistical parameters of forensic and population genetic interest will be calculated by STRidER from the validated data and may serve as the basis for publications and population comparisons. All formulae will be specified and referenced. Therefore, STRidER can set standards of accuracy and appropriate transparent statistical analyses for STR data. A unique, permanent, and citable accession number will be connected to every dataset and related publications will be listed. STRidER will be continuously curated, and a release history on the website will announce additions and amendments.

4. Outlook: digging deeper into variation – STR sequence data in STRidER

Variation at STR loci is currently almost exclusively assessed from electrophoretic size-based categories and interpreted as repeat numbers. The long-recognized sequence variation in repeat units and flanking regions [4,5,44,45] was hitherto only rarely studied at the population level (e.g., [46]) until massively parallel sequencing techniques (MPS) recently provided access to this information (e.g., [47–52]). It was demonstrated that sequencing of STRs increases the overall discrimination power compared to that of electrophoretic sizing and offers additional advantages to forensic human identification (summarized in [53]). A uniform allele nomenclature (proposed in, e.g., [51]) has not yet been established [44], but considerations on the implementation of STR sequence data into practical forensic work are being discussed [53]. The storage of nucleotide strings (text strings) in FASTA-like format (see [54]) has been identified as the core framework to permit software-aided alignment and translation between STR nomenclatures. The existing architecture of STRidER allows for the implementation of nucleotide sequence strings (as kept in EMPOP [54]) and thus is fully compatible with the QC of population data generated by MPS. Converting sequences into the conventional electrophoretic-based STR nomenclature allows for backward

compatibility of data that is a key requirement in forensic genetics [53] and implements the suggested “combined search approach” [44], i.e. a length-based search against the entire database and a sequence-based search against the MPS data subset.

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