DNA profiling

DNA profiling is a valuable forensic approach: it can generate investigative leads by retrieving suspect names from DNA databases and informative weights of evidence when persons of interest and evidential traces are compared. The growing number of markers in profiling systems makes DNA profile comparison increasingly time-consuming and error-prone. Software tools may be a solution for these issues.

DNAxs

The Netherlands Forensic Institute (NFI) has developed a software expert system, *DNAxs*, for case data management, within-case matching of profiles and complex DNA profile interpretation by, among other things, providing summary statistics, major donor inference, weight of evidence calculations and enabling DNA database searching. All from within one software system.

DNAxs is a user-friendly, well-validated software that compares profiles within seconds and is able to use statistical analysis according to advanced probabilistic genotyping models. This increases the quality and number of cases that can be handled and stimulates the use of such advanced statistics. Forensic experts can adopt a well-structured and uniform workflow and focus on the scientific aspects of the work by which the overall quality and uniformity of forensic casework will increase. The platform aids the forensic DNA experts in casework by giving overview and managing the increasingly complex data interpretation and decision making process. In addition, DNAxs has increased consistency and accountability, while reducing errors and interpretation variability[1].

DNAStatistX

Within the software, the DNAStatistX module integrates the continuous maximum likelihood ratio model based on EuroForMix (www.euroformix.com) to calculate the evidential value with probabilistic DNA statistics effortlessly[2].

Software Testing

To ensure that the software is robust and behaves as designed, the DNAxs software is tested during development, prior to release and during validation. Automated tests were designed and built for DNAxs. These tests include unit tests and integration tests. In addition, prior to release for use in forensic casework acceptance testing is performed in which functionalities are tested manually.

Software Validation

DNAxs, including the DNAStatistX module, are NFI-validated according to internal procedures in line with the ISO 17025 standard. Next to that the software was subjected to a multi-laboratory validation study in which four laboratories participated: the Netherlands Forensic Institute in the Netherlands, Institute of Legal Medicine in Austria, National Forensic Laboratory in Slovenia, Institute of Legal Medicine (Cologne) in Germany, and Institut National de Police Scientifique (Ecully) in France. The study was partly funded by the European Union's Internal Security Fund — Police (Proposal Number: 820838, Proposal Acronym: DNAxs2.0). Overall, the software was found intuitive, user-friendly and valid for use in multiple laboratories[3].

[1] Slagter et al. The DNAxs software suite: A three-year retrospective study on the development, architecture, testing and implementation in forensic casework. Forensic Sci. Int. Reports (2021).
[2] Benschop et al. DNAxs/DNAStatistX: Development and validation of a software suite for the data management and probabilistic interpretation of DNA profiles. Forensic Sci. Int. Genet. 42 (2019) 81-89.
[3] Benschop et al. Multi-laboratory validation of DNAxs including the statistical library DNAStatistX. Forensic Sci. Int. Genet. 49 (2020) 102390.

Software functionalities summarized

Functionalities		Details
Profile	Profile detail view	• Bar graph representation of detected alleles per locus and corresponding quantity (peak heights or read counts). • Combine multiple runs into one profile. • Main contributor/ consensus/ composite. • Change locus order to different autosomal STR kits. • Scale Y-axes per locus or per profile. • Set stochastic threshold. • Compare profile with other profiles in the case. • View EPG when accessible in PDF.
	Profile info	• Automatically derive complexity, quality, duplicates and gender. • Total allele count (TAC). • Maximum allele count (MAC). • Numbers of Type I, II and III loci (according to LoCIM [2]). • Number of contributors estimation. • Remaining alleles (after comparison with other profile).
	Profile functionalities	• Create/ infer unknown person (LoCIM/ deconvolution/ manually). • Edit or deactivate alleles, loci or runs.
	Trace similarity	• Automatically matches traces with other traces in the case.
	Sorting profiles	• Rename/ tag profiles.
Tools	Match matrix	• Match matrix enabling comparisons of (many) stains to (many) reference profiles or other stains. • Minimum number of non-matching alleles between the profiles. • Colour coding for the number of non-matching alleles and a slider to set this number. • The match matrix can be exported to PDF. • Profile comparison can be viewed in detail by opening the EPGs or profile details.

Weight of evidence	Maximum Likelihood (ML)	• Create ML hypothesis.
		• View ML calculation results
		  ○ Result table
		  ○ Validation
		  ○ Optimizer steps

Searches	SmartRank	• Create SmartRank [8,9] hypotheses. • Search selected databases (elimination, criminal). • View SmartRank results. • Add candidate retrieved from SmartRank search to case.
Import	Autosomal profile (CE based)	• Case number, ProfileID and RunID can be retrieved from SampleID using sampled pattern. • Comma Separated Value files (CSV).
Export	Autosomal profile	• Comma Separated Value (CSV). • Familias (CSV) [15]. • Bonaparte (via API) [16]. • LIMS xml.
Audit trail		• Keeps track of which actions are performed when and by whom.
Notes		• Add notes to a case. • Add notes to a profile.

Software integration

The software can link to other software tools such as

- SmartRank (https://github.com/smartrank/smartrank)
- Bonaparte (https://www.bonaparte-dvi.com)
- FDSTools (https://www.fdstools.nl/)
- CODIS (https://www.fbi.gov/services/laboratory/biometric-analysis/codis).
- LIMS systems

Software installation

DNAxs can be installed as a stand-alone version on a personal computer or (as a more complex solution) installed on a Linux server which can be accessed by multiple users.

Hardware/software Requirements

System requirements:

- Minimum memory: 4 GB
- Recommended memory: 8 GB for Windows platforms, 6 GB for non-Windows platforms
- Minimum disk space: 10 GB
- Recommended disk space: 50 GB
- 2-4 CPU cores; 4 minimum if statistical calculations on four-person mixed DNA profiles are desired

Software requirements:

- Java 1.8 or higher

Supported browsers:

- Microsoft Edge
- Google Chrome
- Mozilla Firefox

Software availability

A free version of the software (DNAxs2.0) is available at request (dnaxs@nfi.nl) and can be used until the end of 2021. A supported and updated version of the software with all functionalities is available at three different license levels. The annual fee (2021) for the supported version includes both support hours and hours to cover the general development of the software.

	1-2 users	3-6 users	> 6 users
Hours total	11	21	36
Hours support	7	14	25
Hours general development software	4	7	11
Total tariff	**€ 1.816**	**€ 3.514**	**€ 6.098**

More information

For more information visit https://www.forensicinstitute.nl/research-and-innovation/international-projects/dnaxs or send an email to dnaxs@nfi.nl

DNAxs screenshots

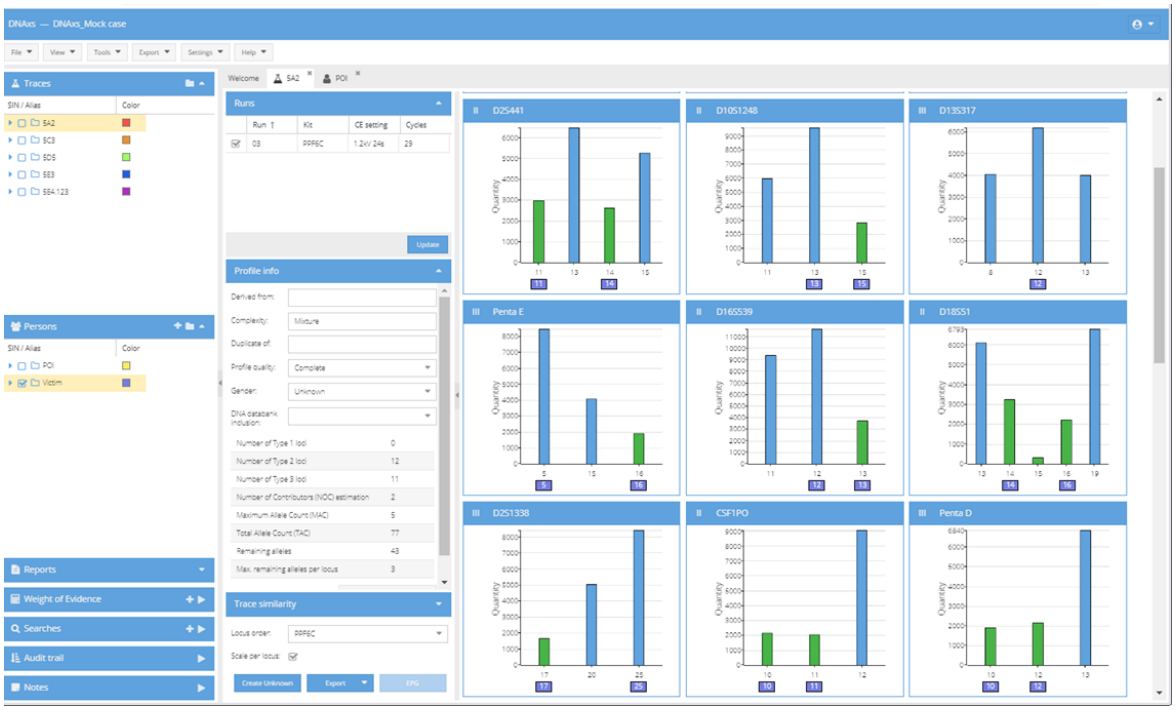

Figure 1. Screenshot of a case in DNAxs with five traces and two reference profiles. The bar graphs for the first trace are presented. The reference profile of the victim is matched underneath the bars of the trace profile.

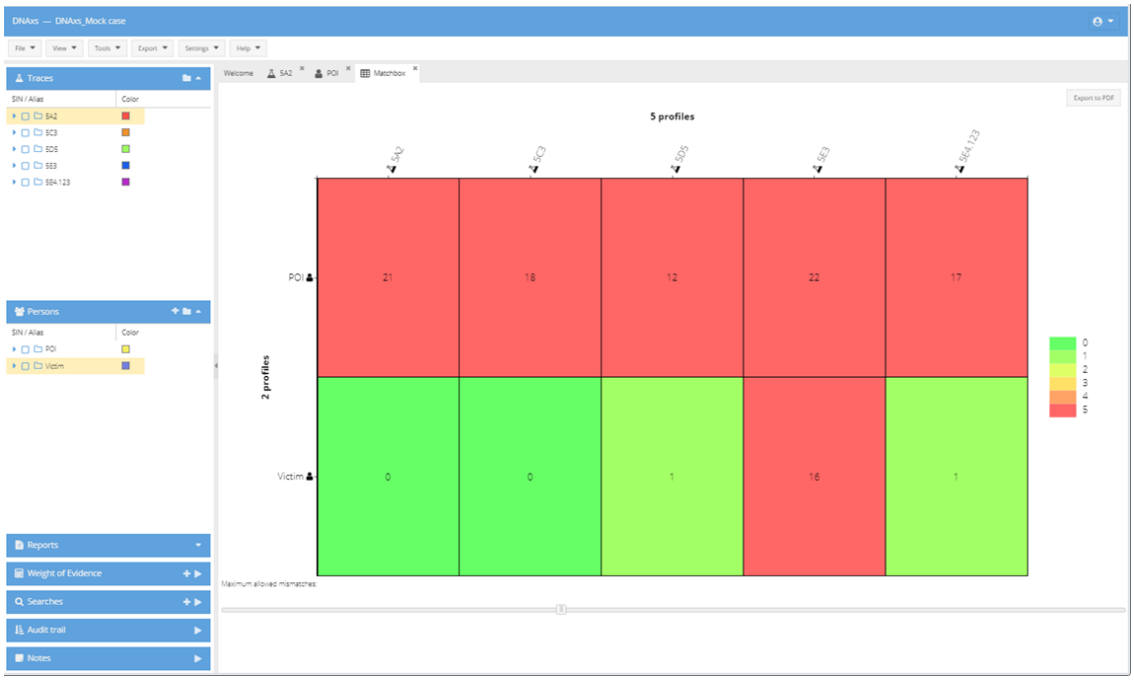

Figure 2. Match matrix comparing the five traces (horizontally) to the two reference profiles (vertically) in a case. Increasing numbers of non-matching alleles are presented as colors ranging from green via orange to red. The actual number of unseen alleles is presented per comparison.

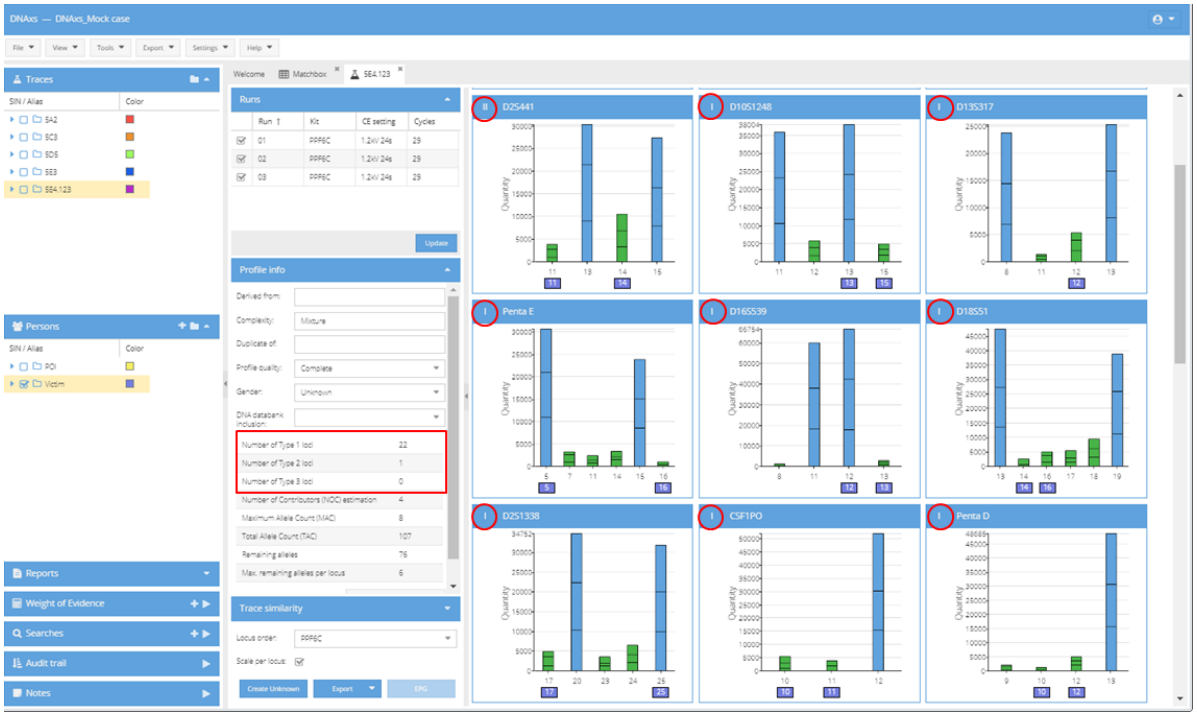

Figure 3. DNAxs bar graphs of a trace that includes three replicates. The red-outlined square shows the total numbers of Type I, II or II loci as determined by LoCIM enabling inference of the major component. The locus type per locus is encircled in red. Blue bars indicate the alleles that are inferred by LoCIM as being from the most prominent component of the mixed DNA profile.

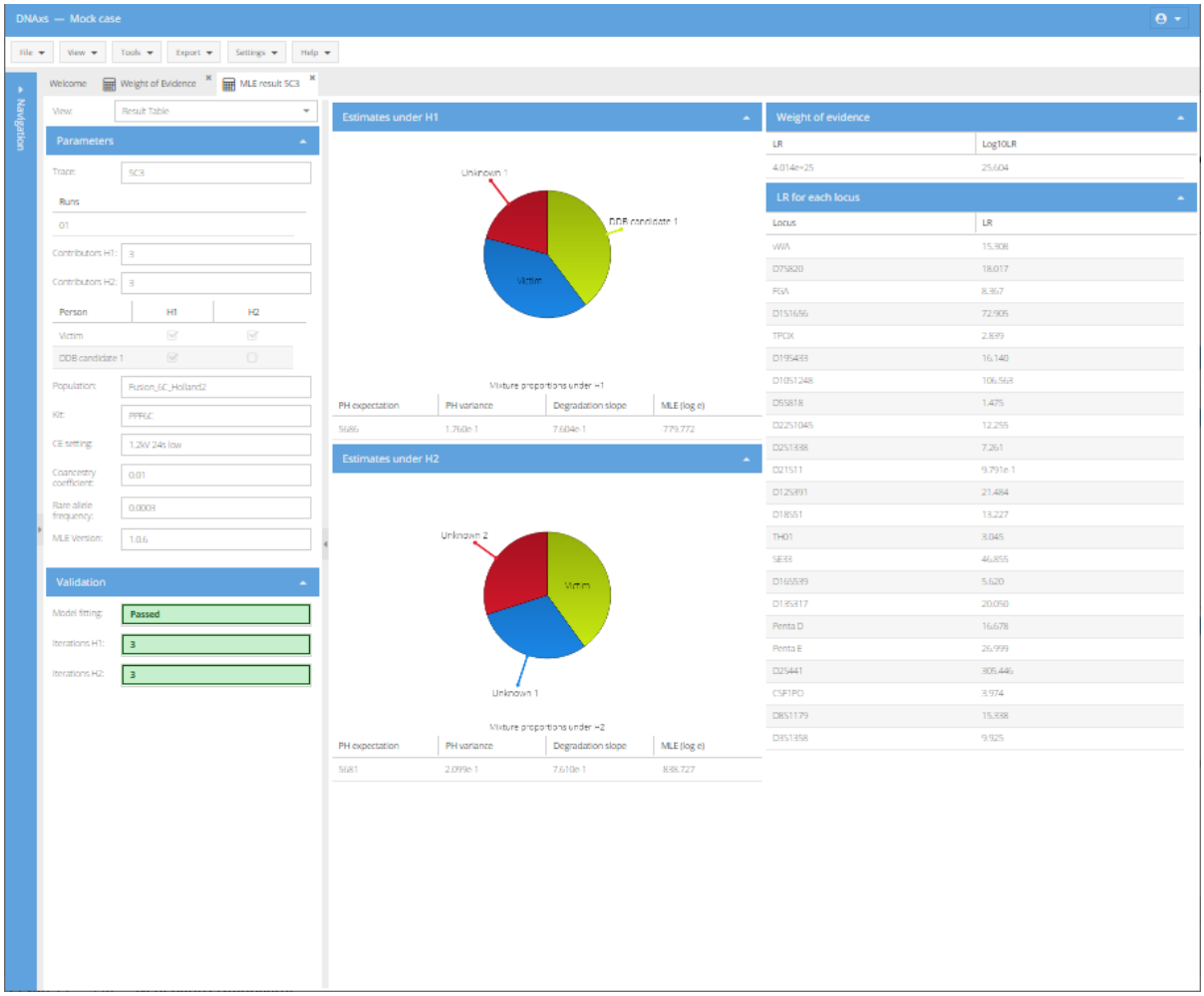

Figure 4. Screenshot of DNAStatistX Result Table within DNAxs. An LR was calculated using a three-person mixture and a person of interest (denoted 'DDB candidate 1'). The results table presents the chosen parameter values, the model estimated parameter values, the overall LR and LRs per locus, and the model validation and optimizer iteration results. Further details can be examined when selecting a different view option (i.e. Model validation or Optimizer).