

# **BONAPARTE : Bayesian networks for** victim identification on the basis of DNA profiles

An ICIS valorisation project proposal SNN, Nijmegen (the proposer and the executive) NFI, The Hague (the costumer)

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## 1 Summary of the project

The objective of the project is to improve NFI's (Netherlands Forensic Institute) missing person screening and matching routine for victim identification based on DNA profiles. A particular aim is to enable NFI to handle the screening and matching task in case of a large national disaster with many unidentified victims. An additional aim is to help ICMP (International Commission on Missing Persons, Bosnia) with methods to improve their kinship analysis system, which is, for instance, used for analysis of data from mass graves in the Balkan and international disasters such as the tsunami in 2004 and the hurricane Katrina in 2005.

One of the tasks of forensic research --such as is performed at the NFI -- is the identification of unidentified victims through screening and matching of their DNA profiles against the DNA profiles from relatives of missing persons. In this task, the quality of the match is obviously of great importance for humanitarian reasons, and, to some extent, economic reasons as well. The matching task is often indirect (if there is no material of the missing person him/herself) and should be performed using statistical methods. Bayesian networks are very well suited to model the statistical relations of genetic material of relatives of the missing persons are used in the screening phase. As a result, with these models correct matches can be found at the cost of much less false hits than with more straightforward methods which do not take pedigree information into account. Another important advantage of a Bayesian network approach is that it makes the analysis tool more transparent and flexible, allowing to incorporate other factors that play a role - such as measurement error probability, missing data, statistics of more advanced genetic markers etc.

The proposed ICIS project *Bonaparte* will be part of NFI's *Napoleon* project. Napoleon's aim is to develop Bayesian network methods and software for the NFI for the identification of unidentified victims through screening and matching of their DNA profiles against databases with DNA profiles of relatives from missing persons. Bonaparte will be the technical core in Napoleon. The Bonaparte project will research, design and develop flexible and transparent models using automatically generated Bayesian networks and Bayesian inference methods, enabling to correctly do kinship analysis on the basis of DNA profiles combined with pedigree information. In addition, the Bonaparte project will develop a graphical user interface for forensic analysts. Means for communication with other NFI systems (e.g. for the DNA-data input) will also be implemented. These will be generic, such that Bonaparte can be coupled to systems in e.g. other forensic institutes such as ICMP as well.

The project will have two phases. Assuming start data 1-3-2008, in phase 1 the basic functionality will be implemented, to be finished in 12-2008. Phase 2 is for extended and advanced functionalities, to be finished in 10-2009.

## 2 Valorization checklist

#### 2.1 Project participants and organization

- Who will lead the *project*? Which companies and/or organizations (universities, industries, institutes) will participate? For which percentage of the effort?
  - SNN will lead Bonaparte and is the only participator
  - Bonaparte is a subproject of the larger Napoleon project of NFI. Effort in Bonaparte is 57% of the total Napoleon effort. Napoleon is lead by NFI. NFI and its subcontractors (IT companies) will carry out the other 43% of Napoleon.
- Do any of the *project* participants already participate in ICIS?
- SNN already participates in ICIS.
- How will the *project* be run or embedded? For example by a consortium/proposer, within the ICIS I&D or other cluster, within an end user organization.
  - The Bonaparte project will be run as part of the Napoleon project lead by NFI.
    NFI will have other subcontractors in the Napoleon project as well. NFI and ICMP will be involved as end-user.
  - ICMP = International Commission on Missing Persons ("Bosnia", "tsunami", "Katrina"), see section 3.4

#### 2.2 Customer and end users

- Who is the customer of the ICIS *contribution* such as ICIS knowledge or technology?
  NFI is the customer of the ICIS *contribution*.
- Is there an end user or branch association involved in the project?
- NFI and ICMP will be involved as end-user in the project.
- Is there any demand articulation activity foreseen or necessary?
  - There is a demand for effective, flexible, efficient and transparent software that can do large scale DNA matching of unidentified victims with a database of relatives of missing persons, in particular in case of disaster, where many data has to be handled. Currently available systems are considered inadequate by end-users.

#### 2.3 Innovation and customer benefits of the project results

- Which aspects of innovation are targeted by the *project* and/or *the contribution*? For example 'newness' (new to the world, new for a market, new for a company) and 'innovation impact' ('schudt de markt wakker' or incremental improvement)
  - The resulting product will be new to NFI and ICMP ,and to other international forensic institutes as well. Important improvements are
    - That forensic institutes can handle more data of missing persons more effectively, with much less false positives and much less manual postprocessing
    - The flexibility and transparency of the system due to the use of Bayesian networks
- What are the more specific goals/objectives of the *project* and *the contribution*?
  - The objective of the project is to improve NFI's unidentified victims-missing persons screening and matching routine. An important objective in here is to enable NFI to handle the screening and matching task in case of a national

disaster with many unidentified victims. A third aim is to improve ICMP's unidentified victims-missing persons screening and matching systems.

- What will be the social/economic effects of the *project*? For whom?
  - Social benefits: relatives of victims/missing persons will be helped better and faster. In particular in a disaster scenario this will have much social impact, since the identification of so many victims is almost impossible without an effective automated system. Who: relatives of victims of a national disaster (NFI), relatives of victims of international disaster and armed conflicts (ICMP).
  - Economic benefits: a more efficient routine at the NFI.

### 2.4 Utilization of ICIS assets

- What ICIS contributions will be utilized in this *project*? For example specific knowledge/technology assets previously developed in ICIS.
  - Automated Bayesian network modeling and efficient inference.
  - What problem(s) will be (re)solved using these assets?
    - The problem of how to deal with large scale databases of DNA from victims and relatives of missing persons such as will occur in case of large disasters and the problem of many false hits that occur in current DNA kinships analysis on such data.
- Any spin-of research foreseen?
  - Implementation of Bonaparte at ICMP or other international institutes is expected to generate further demand for technological research and development.

#### 2.5 What will be the outcomes of the valorisation?

- How will the *contribution* (utilization of ICIS assets) materialize in the project? For example transfer of knowledge, embedded in a proof of principle or in a product)
  - The contribution will materialize as transfer of knowledge of Bayesian network methods for victim identification and a generic technological core in the Napoleon project.
- How will the *contribution* help to embed (verankeren) ICIS results?
  - The contribution will have a central role in a relevant application that will be used by the renowned NFI forensic institute and is foreseen to play a central role in ICMP and other international forensic institutes as well.
- Are there any separate/identifiable *project* deliverables that will contribute to the ICIS goals? Will the project contribute to the common ICIS demonstrator? How?
  - This project deliverable will be important for crisis management in national and international disaster scenarios, and in this way, the project will contribute to the common ICIS demonstrator.
- Are any patents or other type of IP foreseen? Will ICIS partners share is the revenues?
  O IP will remain at SNN.

#### 2.6 Dissemination

- The project will deliver a publication. • Yes
- The project can contribute to the Scientific Book or Glossy Booklet. Will the project contribute to the Scientific Book or the Glossy Booklet?
  Yes
- The project can contribute to special events.

o Yes

#### 2.7 **Costs and financial ICIS contribution**

- What are the (minimal) total costs of the project?
  - Costs of Bonaparte: 10, 1, c & 10, 2, b 0
  - Napoleon's specific costs for NFI developments outside Bonaparte: 10, 2, b 0
  - Total costs Napoleon: 10, 1, c & 10, 2, b 0
- How much subsidy is requested out of the ICIS budget? What percentage of the total cost . are ICIS partner costs? (This part is eligible for ICIS subsidy)
  - o ICIS requested subsidy €130.000,=
  - SNN (=ICIS partner) costs is 100% of Bonaparte 0
  - SNN costs is 57% of Napoleon. 0
- What percentage of the total project costs will be paid for by a customer or 3-rd party?
  - NFI will pay 10, 2, b of Bonaparte
  - This is about 50% of Bonaparte
  - NFI will pay 10, 2, b 0 of Napoleon
    - This is about 72% of Napoleon

Effort by, cost to be paid to:			Cost to be paid by:			
Project	Subproject	Performer/ creditor	NFI 10, 2, b	SNN	ICIS subsidy	Total
Napoleon		NFI		10, 1, c	-	10, 2, b
	Bonaparte	SNN			€130.000,=	
Total Napoleon		and the second	-		€130.000,=	

(\*\*) costs based on a rate o

0

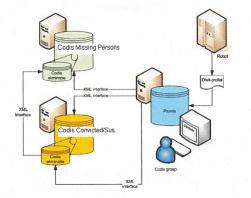
All cost are excl. VAT.

## **3** The project: background and objectives

Within the Napoleon research project the aim of the ICIS valorization project Bonaparte is to research the application of Bayesian network methods to victim identification in which whole pedigrees of the relatives of missing persons are used in the screening phase An important advantage of the Bayesian network approach is that the number of false hits will be much lower than conventional methods. Another important beneficial feature of Bayesian networks is that they offer a mathematically precise, powerful, transparent and flexible modeling approach in which model extensions are easily incorporated.

New flexible and transparent generic software will be developed based on these methods. The software will consist of data-interfaces, a graphical user interface and a screening and matching functionality. Bonaparte's Bayesian network methods for matching of missing persons against its pedigree's in large databases will enable the Napoleon system to improve the quality and continuity of the services delivered by the NFI, even in case of a major disaster. The combination of both the pedigree approach and the Bayesian approach is a unique method in DNA identification work.

#### 3.1 Background



3.1.1 Regular Forensic Cases - current situation

The NFI uses the database CODIS (Combined DNA Index System) in combination with the kinship analysis program DNAView in DNA identification work. CODIS is designed to identify criminal suspects from DNA registers and is designed for direct comparisons of DNA profiles. For regular forensic work CODIS works fine, as designed. But when it comes to identifying unidentified persons through a database of DNA profiles of relatives of missing Persons (indirect comparisons), it runs short,

which will in particular a problem in the case of a large disaster. The kinship analysis program DNAView cannot use whole pedigrees with DNA profiles from the relatives of missing persons in the screening phase, it gives many false hits.

#### 3.1.2 Catastrophes

#### World Trade Center in New York disaster on 11 September 2001

It is now estimated that 2,749 lives were lost in the terrorist attacks on the World Trade Center in New York on 11 September 2001. The software used in the first weeks after the disaster, CODIS, ran short in this particular case. For identification of the victims of this disaster the program M-FISys (Mass-Fatality Identification System) has been developed by Gen Codes Forensics. This software is very expensive, and not very transparent. Users of the software are dependent on the maintenance and support of Gen Codes Forensics. Outside the US, only the Metropolitan Police in England has purchased the software, but their experience so far is not positive. Even the ICMP, an institute at which a lot of identification work is performed, has not purchased this M-FISys software.

#### The East Asia Boxing Day tsunami disaster

The East Asia Boxing Day tsunami disaster presented different challenges. Although few of the bodies were fragmented, the disaster was on a far larger scale and, with many members of some families being lost; proportionally fewer kinship records were available. It was sometimes possible to identify someone from records from a relative who was also a victim. A disaster on this scale also gives rise to an identification problem of a higher magnitude again.

#### 3.2 Objectives of Napoleon

The main objective of the Napoleon project is to provide the NFI researchers and analysts an efficient and effective tool, which can help in the identification of unidentified persons through screening and matching of unidentified person's DNA profiles against the pedigrees of relatives from missing person's DNA profiles in large databases. In this way the Napoleon system will improve the quality and continuity of the services delivered by the NFI, and it should be able to handle data of a major disaster.

The basic productive system should be ready by 31-12-2008. A full and advanced production system that can handle complex pedigrees and partial DNA profiles should be available by 01-10-2009.

#### 3.3 Objectives of Bonaparte

For efficiency, transparency and flexibility purposes Napoleon's core module will be based on a Bayesian network analysis approach. This will be researched and developed in the Bonaparte project as ICIS valorization. The Bayesian network approach will significantly reduce the number of false hits, and thus streamline the process of identifying victims. In addition, it allows that (future) model extensions are easily facilitated, such as

- Measurement errors
- Mutations
- Population structure
- Missing data
- Advanced genetic markers (SNP's, LCN's)
- Alternative hypothesis testing

#### 3.4 ICMP: International Commission on Missing Persons

The International Commission on Missing Persons (ICMP, <u>www.ic-mp.org</u>) was established at the initiative of U.S. President Clinton in 1996 at the G-7 Summit in Lyon, France. Its primary role is to ensure the cooperation of governments in locating and identifying those who have disappeared during armed conflict or as a result of human rights violations. ICMP has its headquarter and research lab in Bosnia. ICMP is responsible for identifications of victims in Bosnia, after Tsunami disaster, Katrina hurricane and after other disasters. Since its establishment in 1996 ICMP has been funded through voluntary grants, donations and contributions from participating governments and organizations.

NFI will incorporate the pedigree-functionality into the ICMP-system at no cost for ICMP . SNN will grant the ICMP to use this algorithm without license fees under specific conditions.

NFI researchers explicitly expressed to consider it to be an honor to support and work together with the ICMP on its humanitarian mission. We sympathize with this idea and will therefore cooperate with NFI in this issue. In our opinion this will suit very well in the context of the ICIS project, using public money to valorize technology for the benefit of victims of armed conflicts and disasters. Beside this, it will be a perfect opportunity for ICIS publicity.

## 4 The contribution: description of the proposed system

The Bonaparte system will be based on a Bayesian network analysis approach. It is well known that Bayesian networks are well suited for genetic analysis involving complex inheritance models and family data. The reason is that the biology of genetic inheritance in pedigrees leads to statistical models which are naturally described as Bayesian networks.

The modularity and transparency of Bayesian networks allows models to be easily adapted to specific parameter settings such as e.g. allele mistyping probabilities and mutation rates. In addition, models can be extended to incorporate other sources of uncertainty, such as the uncertainty whether a certain object (e.g. a toothbrush) is indeed the object belonging to the missing person (and not to his brother), or the uncertainty whether a supposed father is indeed the biological father.

Once a Bayesian network has been specified, highly efficient inference algorithms can be applied to calculate the statistics of interest. The inference algorithm is general and does not need to be adapted to a particular model. This separation of the modeling stage and the calculation stage is an essential advantage of using Bayesian networks and increases efficiency of the entire procedure.

Publications by members of our group (see below) and other groups (Kong, 1991; Fishelson and Geiger, 2002; Lauritzen and Sheehan, 2003) have demonstrated the usefulness of Bayesian networks in the context of genetic analysis.

#### 4.1 R& D, modelling

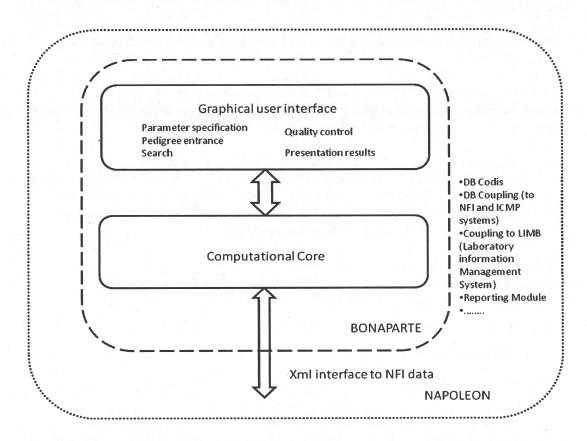
These properties make Bayesian networks the tool of choice for forensic analysis for victim identification based on DNA profiles. How exactly Bayesian networks are to be modeled in this specific application field, including all the desiderata (see appendix I), such that the tool can handle many data automatically without putting too much modeling burden on the forensic researcher is part of the research and development part of the Bonaparte project. Results of this part will be generic methodology. These will be reported and published. The implementation in the Bonaparte module is described in the following section.

#### 4.2 System development

The Bonaparte system that we propose to contribute will be part of the Napoleon system. The system will have two components (see figure below):

- Computational core (Bayesian modeling and inference)
- Graphical User Interface

The system interacts with the other NFI systems, such as databases, via XML protocols.



4.2.1 Operation of the system

The system will operate as follows:

Relevant data needed for the kinship analysis (victims' DNA data, DNA data from objects and relatives and possibly relations in families of missing persons) will be imported from other NFI systems into the system via xml protocols. On the basis of these data supplemented with specifications (parameters, family structures) provided by the forensic analyst via the GUI, the computational core generates a series of Bayesian networks. With these networks likelihoods for the matches that are to be screened. Results are presented via the GUI and can be communicated to other NFI systems via XML. The XML interface will be generic, such that Bonaparte can be coupled to systems in other forensic institutes (e.g. ICMP) as well.

The system can be controlled via the GUI: configuration of parameters, inspection of results, quality control of data, options to provide the system with alternative hypotheses, search commands, and presentation of results. Furthermore, the system will have features such as the inclusion of annotations, authorization of matches, and history of analysis.

The system will be implemented as a multi-client system. It should be able to run on a standard laptop.

#### 4.2.2 Embedding of the system within NFI

Bonaparte will be a subproject of Napoleon. Other parts of the Napoleon project will be carried out by NFI and/or subcontractors. These parts include the further embedding and integration of the system with other NFI systems, databases, and processes, automatic reporting module, specification of the end-user part of the GUI etc. These are not further discussed here. Also integration with ICMP systems and processes will fall under other parts of Napoleon, not discussed here. For a detailed description of the functionality of Napoleon system we refer to Appendix I.

#### 4.2.3 Intellectual property rights and licenses

Intellectual property (IP) rights of Bonaparte remain with SNN. NFI will obtain a nonexclusive license. License costs for NFI are covered by NFI's contribution to Bonaparte. ICMP will be granted a non-exclusive license free of charge under conditions to be agreed. ICIS community will obtain a non-exclusive license allowing to use Bonaparte for scientific research and demonstration purposes within the ICIS project.

## 5 Time schedule, effort and budget

### 5.1 Time schedule

The proposed project will be carried out in 2 phases. In the first phase, we will research and develop the basic functionalities. In the second phase, we will develop additional functionality, such as the clustering of partial DNA profiles, and the handling of more advanced DNA markers. NFI strongly requested that the first phase is finished by the end of 2008 and the second phase by 10-2009. This is realizable under assumption of start date 1-3-2008.

Phase	Start date	End date
1	1-3-2008	31-12-2008
2	1-1-2009	1-10-2009

### 5.2 Effort

Task	Person-months		
	Phase 1	Phase 2	
R&D, Modeling, design	7	4	
Implementation, testing	7	4	
Manual, reports	1	1.5	
Project management	1	0.5	
Total	16	12	
Total project		28	

These estimate are partly based on our experience in the SHELL ICIS valorization project. We satisfactory completed the SHELL project in about 20 person-months, and by comparing innovation and complexity levels, we can get an estimate for the required effort in this project. Our estimate is that phase 1 is a bit less innovative but somewhat more complex than the Shell project. However in the SHELL project, the overall approach to the problem was initially less clear than that we expect in this project. This required a few months extra in the SHELL project. Thus we think that in this project 16 months is a fair estimate. Phase 2 will be more complex than phase 1, but we can build to a large extend on the results and experiences of phase 1.

#### 5.3 Budget

For application projects, SNN charges standard a commercial rate of 10, 2, 6 uro/day. Budgets are computed assuming 20 days per month.

The total effort is 28 months. The budget for Bonaparte is therefore 10, 1, c & 10, 2, b SNN is willing to invest 10, 1, c in Bonaparte. ICIS subsidy of €130.000,= is requested. If subsidy is granted, the remaining 10, 1, c & 10, 2, b or the Bonaparte project is payable by NFI to SNN.

The additional NFI effort to Napoleon is estimated to be 3292 hours, partly to be carried out by NFI and partly outsourced by NFI to other parties. For these hours, NFI calculates a rate ot 10, 2, b hour. So, the additional NFI efforts are estimated to be 10, 2, b he total budget of the Napoleon project is the sum of the Bonaparte budget of 10, 2, b and the additional effort in Napoleon, 10, 2, b , which equals a total of e

The requested ICIS subsidy is about 15% of the total Napoleon budget, which corresponds to a valorization part of about 30% of total Napoleon budget.

Effort by, cost to be paid to:			Cost to be	paid by:		
Project	Subproject	Performer/ creditor	NFI 10, 2, b	SNN	ICIS subsidy	Total
Napoleon		NFI	_1U, 2, D	<b>†</b>	-	10, 1, c 10, 2, b
-	Bonaparte	SNN			€130.000,=	
Total Napoleon					€130.000,=	

(\*) costs based on rate of  $\begin{pmatrix} 10, 1, c \\ 10, 2, b \end{pmatrix}$ (\*\*) costs based on rate o

All cost are excl. VAT.

#### **Project team** 6

#### 6.1 **SNN Team**

For phase 1, we will have about 10 months time for a 16 person-month project. So we need a project team with about 2 persons per tasks to carry out the project. This project team will consists of the following people.

- 10, 2, e Modeling:
- 10, 2, e Programming
- 10, 2, e Manual and reports:
- 10, 2, e Project management: 10, 2, e

#### 6.2 **Experience of SNN team**

Drs 10, 2, 6 has been responsible for the succesful software implementation in valorization project with SHELL. " A Petronhysical Decision Support System". In this project, <sup>10,2,e</sup> 10,2,e

cted as supervisors/advisors.

- 10, 2, e has been responsible for the BayesBuilder JAVA implementation (ICIS-CDM), supervised by <sup>10, 2, e</sup> and the JAVA implementation of Promedas, a system applying Bayesian networks for medical diagnosis (supervised by 10, 2, e
- Drs 10, 2, e has performed his PhD research at SNN in the field of genetic applications of Bayesian networks<sup>10, 2, e</sup> acted as his advisors.
- Dr<sup>10, 2, e</sup> s researcher at SNN in the field of Bayesian networks. He also carried out and supervised research in genetic applications of Bayesian networks. He has ample experience with database application in genetic anthropology. Currently he is director of Promedas BV, applying Bayesian networks for medical diagnosis.
- DI <sup>10, 2, e</sup> s researcher at SNN in the field of Bayesian networks. He also carried out research within the ICIS-CDM, and has been supervised other ICIS projects. Currently he is vice-director of Smart-Research BV, for commercial applications of neural networks and Bayesian networks.
- Prof Dr <sup>10, 2, e</sup> is professor "Neurale netwerken en machine intelligentie" . and director of SNN. He supervised many projects, including ICIS projects.

#### 6.3 **Relevant publications of SNN team**

Albers CA, Stankovich J, Thomson T, Bahlo M, Kappen HJ (2008) Multipoint approximations of identity-by-descent probabilities for accurate linkage analysis of distantly-related individuals American Journal of Human Genetics (in press)

Albers CA, Heskes T, Kappen HJ (2007) Haplotype inference in general pedigrees using the cluster variation method Genetics 177:1101-16

Albers CA, Leisink MA, Kappen HJ (2006) The cluster variation method for efficient linkage analysis on extended pedigrees BMC Bioinformatics 7(Suppl 1):S1

Albers CA, Kappen HJ (2007) Modeling linkage disequilibrium in exact linkage computations: a comparison of first-order Markov approaches and the clustered-markers approach BMC Proceedings 2007, 1(Suppl 1):S159

de Vries BB, Pfundt R, Leisink M, Koolen DA, Vissers LE, Janssen IM, Reijmersdal S, Nillesen WM, Huys EH, Leeuw N, Smeets D, Sistermans EA, Feuth T, van Ravenswaaij-Arts CM, van Kessel AG, Schoenmakers EF, Brunner HG, Veltman JA (2005) Diagnostic genome profiling in mental retardation. Am J Hum Genet. 2005 Oct;77(4):606-16. Epub 2005 Aug 30.

### 6.4 NFI team

The following persons from NFI are involved in the Napoleon project from which the ICIS project Bonaparte will be a subproject.

- Executive Napoleon <sup>10, 2, e</sup> Head of department BSO "Biologisch Sporen Onderzoek") <sup>10, 20, e</sup>
- End-user Napoleon, forensic scientist, specialist kinship analysis: Dr

		project	leader	Napoleon:	10, 2, e 1
10	2 e				1 3 5 5 5 7 7 7

#### 6.5 ICMP team

The following persons from ICMP are involved in the Napoleon project.

•	10, 2, e	ICMP Bosnia	10, 2, e
•		ICMP Bosnia <	

### Appendix I Napoleon functionality

As defined in collaboration with NFI (including the non-ICIS parts) **Basic Functionality** 

- Operation in a network environment with multiple users, or as a standalone system that can be run entirely on a common laptop.
- Databases with DNA-STR profiles configuration and maintenance Add database; Delete database; Convert old database to new database. [With database-creation and ETL scripts]; Import DNA profiles [ per group/cluster?]; Export DNA profiles. [ per group/cluster?]
- Static Data configuration and maintenance: Population STR data (frequencies): Adding; Search; Update; Delete Locus table: adding/update/delete locus Amplification Kit table: adding amplification kit loci specifics Maximal 25 STR Data compatibility with all current commercially available kits and markers and extensible for custom systems and marker sets as needed, to ensure long-term usability
- Static Data configuration and maintenance: Population mutation data for STRs: Adding; Search; Update; Delete
- DNA profile tables (authorized) modifications [per group/cluster?] Add loci, alleles, unique DNA identity number, casenumber Search, Update, Delete
- Option to compatible file exchange with CODIS
- Automatic screening of all samples against Elimination Known profiles (e.g., laboratory personnel), reducing the chance of misidentification through laboratory contamination of samples: could be performed by CODIS itself
- User (Role Based) Access Control

**DBA** function

Multiple levels of secure access privileges including "Administrator Only" functions. Specific operators can be assigned to samples or groups of samples

- Pedigree: Kinship (table) configuration: Adding; Search; Update; Delete
- Matching options: options are optional and can be configured by the user [defaults to default values]
  - option to select DNA profiles and pedigrees for searching
  - option to select a population for statistics
  - option to select a subpopulation correction
  - option to chose a frequency for rare alleles
  - option to add the maximal number of mutation loci in a case
  - option to include null alleles in match
  - option to include mutation model of K. Ayres and C. Brenner
  - option to give the minimum required loci in match
  - option to add a prior probability

- option to calculate beside LR, the posterior probability

- option to chose thresholds for reporting results
- Direct searches:

One-to-many and many-to-many searches between DNA profiles of Missing Persons against DNA profiles of Unidentified Persons

*One-to-many* and *many-to-many* searches between DNA profiles of Unidentified Persons (/bodies parts) against DNA profiles of Unidentified Persons (/bodies parts) in order to group profiles

- Indirect searches: One pedigree-to-many-profiles and many pedigrees-to-many-profiles searches between DNA profiles of Reference Samples against DNA profiles of Unidentified Persons
- Automatic Calculation of likelihood ratios and posterior probabilities with flexible, intuitive parameters based on Bayesian Stochastic Network Analysis. Results are ranked and results above a selected threshold are reported
- Kinship matching to family references. Samples can contribute to more than one case and Napoleon can handle complex pedigrees. Profiles can have multiple roles, allowing a direct reference or newly identified person to be used as a reference for another. Profiles can be reassigned to other references if needed, greatly increasing their flexibility
- Generate user specified Reports from Matching-Results-table Report options: searching defaults (see matching options) Print Reports; Save Reports

Export matching results from Matching-Results-table

- Reassignment of DNA profiles of missing persons found matching
- QA tools allowing you to spot inconsistencies and errors in the data that may be the result of commingled remains, sample switches or contamination and greatly reduce the chance of a false-identification
- User's friendly GUI: Menu's, Options and defaults set up, search and filter commands
- Documentation
- Back-up

#### **Extended functionality:**

Extended functionality will be completely described during phase 1.

- Includes all basic functionality
- Databases with DNA-Y chromosomal STR profiles configuration and maintenance: Adding; Search; Update; Delete; Import; Export
- Static Data configuration and maintenance: population Y-chromosomal STR data:

Adding; Search; Update; Delete Locus table: Adding/Update/Delete locus Amplification Kit table : adding amplification kit loci specifics Maximal 20 Y-chromosomal STR Data compatibility with all current commercially available kits and markers and extensible for custom systems and marker sets as needed, to ensure long-term usability

• DNA Y-chromosomal profile tables (authorized) modifications [per group/cluster?]:

Adding; Search; Update; Delete

Option to compatible file exchange with other external databases (not only CODIS)

- Analyses can be grouped using variable parameters to identify fragments belonging to the same person and to build up a <u>consensus DNA</u> profile from badly damaged remains
- <u>Virtual Profiles</u>, combining multiple test results from a single sample, with concordance & conflict reporting. This greatly simplifies work with fragmented samples, or degraded samples that have been assayed many times
- Matching options: options are optional and can be configured by the user [defaults to default values])
  - option to chose more that one population for the statistics
  - option to include Y chromosomal data for screening
  - option to use fatherless calculation for loci with paternal exclusion
  - option to use motherless calculation for loci with paternal exclusion

#### **Advanced functionality**

Advanced functionality will be completely described during phase 1.

- Includes all extended functionality
- DNA profile tables (authorized) modifications [per group/cluster?] Add: include peak heights; Search; Update; Delete
- Databases with DNA-SNP data and maintenance Add database; Delete database; Convert old database to new database. [With database-creation and ETL scripts]; Import DNA profiles. [per group/cluster?]; Export DNA profiles. [per group/cluster?]
- Static Data configuration and maintenance: population SNP data Adding; Search; Update; Delete Locus table: adding/update/delete locus Amplification Kit table : adding amplification kit loci specifics. Maximal 200 SNP Data compatibility with all current commercially available kits and markers and extensible for custom systems and marker sets as needed, to ensure long-term usability
- DNA profile tables (authorized) modifications [per group/cluster?] Adding; Search; Update; Delete
- Matching options: options are optional and can be configured by the user option to include mtDNA data for screening
  - optionto include SNP data
  - option to use other mutation model (Xu et al 2000)

# Appendix II Glossary of Terms

BSO	(department) 'Biologisch Sporen Onderzoek', department of the Netherlands Forensic Institute.
CODIS	Combined DNA Index System.
ETL	Extracted-Transformed-and-Loaded.
GUI	Graphical User Interface.
ICMP	International Commission of Missing Persons
LCN	Low Copy number. This refers to the situation where there
	are very few DNA molecules available in the sample.
Locus	A position on the genome.
LR	Likelihood Ratio.
M-FISys	Mass-Fatality Identification System, software program
	developed by Gen Codes Forensics.
mtDNA	Mitochondrional DNA.
NFI	Netherlands Forensic Institute.
Null allele	The name implies 'absent' allele. However, in forensic
	work it refers to allele that cannot be visualized. This is
	usually caused by a primer binding site mutation.
PROMIS	Laboratory Information and Mangement System of the NFI.
Rare allele	Allele with a low frequency.
SNP	Single nucleotide polymorphism. Pronounced: "snip".
STR	Short tandem Repeat.
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