

*Approved by Director: Dr. Guy Vallaro*

**14.1 PURPOSE:**

- 14.1.1 To provide guidance for the entry and searching of relatives of missing persons, deduced missing persons, and unidentified human remains in CODIS.

Note: Every effort should be made to obtain multiple family reference samples for each Missing Persons case.

**14.2 RESPONSIBILITY:**

- 14.2.1 DNA Section Personnel.

**14.3 Specimen Completeness Requirements for NDIS and SDIS**

- 14.3.1 STR: Unidentified human remains require one loci and amelogenin for STR data upload at NDIS and SDIS. Missing person samples require seven loci and amelogenin for STR data upload at NDIS and SDIS. Relatives of missing persons require thirteen loci and amelogenin for STR data upload at NDIS and SDIS. Refer to SOP 13.5 and 13.6 for STR data entry and verification.
- 14.3.2 mtDNA: Only DNA data relating to the mtDNA control region shall be uploaded, stored and searched at NDIS. Missing person samples, unidentified human remains, and relatives of missing persons require both HV1 (minimum range 16024-16365) and HV2 (minimum range 73-340) for mtDNA data upload at NDIS. For SDIS, missing persons and relatives of missing persons require both HV1 (minimum range 16024-16365) and HV2 (minimum range 73-340) for mtDNA data upload. Unidentified human remains require a minimum of 200 bases per region (HV1 and HV2) for upload at SDIS with minimum data required from a nuclear DNA profile.
- 14.3.3 Y-STR: If Y-STR data is available, it may be added to the DNA record. In cases where the missing person is a male, Y-STR's should be attempted on a paternal relative.

**14.4 Mitochondrial DNA Data Entry**

- 14.4.1 Launch Analyst Workbench and select Specimen Manager. Select 'mtDNA Data Entry'.

- 14.4.2 Enter the specimen ID (case and item numbers). This information must be entered to continue adding additional details to the profile.
- 14.4.3 Use the tab key to navigate to specimen category and select “other”.
- 14.4.4 Verify the user name (assigned to) is correct.
- 14.4.5 Using the drop down arrow choose source identified as Yes or No.
- 14.4.6 Using the drop down arrow choose yes or no for partial profile. (This is a required field but will default to ‘no’ if not chosen).
- 14.4.7 The alternate lab and alternate specimen ID fields only need to be populated if data is being merged from another laboratory. The NCIC# (National Crime Information Center Number) and VICAP# (Violent Crime Apprehension Program Number) numbers should be populated for all cases. If the numbers are not known, contact the Missing Persons Team at Connecticut State Police – Central District Major Crime Squad (CSP –CDMCS).
- 14.4.8 Add any additional information into the comments field.
- 14.4.9 Right click on the arrow just below the “start” column in the lower half of the screen. This will begin the addition of the mtDNA profile. Click on HV1. The basic information for HV1 will appear on the screen [range (start 16024, end 16365)], read by, and read date. Alternately, the start and end base numbers can be entered manually by clicking in each field and typing the base number. The range entered is the shortest range established between the first analyst and the second analyst of the mtDNA profile data.
- 14.4.10 If the profile being entered has no differences from the rCRS, no more information for HV1 needs to be added. If the profile being entered does have differences, click on the ‘+’ key located next to the arrow. This will expand the HV1 fragment and allow for the entering of additional information for HV1.
- 14.4.11 Click in the empty box below ‘Reading 1: Position’. Enter in the base number of the first difference for the profile you are entering (i.e. if the first difference is 16129 A, enter in the number 16129). Press the tab key to navigate the next field.
- 14.4.12 Enter in the letter of the base difference (i.e. A). Press the tab key to navigate to the next field, ‘Reading 2: Position’.

- 14.4.13 Enter the position number again (i.e. 16129). Press the tab key. Enter the base difference letter again (i.e. A). Press the tab key to navigate to the next difference for sample profile being entered.
- 14.4.14 Repeat the steps 14.4.11- 14.4.13 to continue to add all the differences for HV1.
- 14.4.15 Once data entry for HV1 is completed, right click the '\*' to begin with HV2. Alternately, the user can click in the empty field for the 'start' column and the 'end' column and manually enter the start and end base positions for HV2. The range entered is the shortest range established between the first analyst and the second analyst of the mtDNA profile data. Clicking the '\*' and selecting HV2 will bring up the basic information for HV2 [range (start 73, end 340), read by, and read date]. Repeat the same steps for HV2 that were done for HV1 to add the differences. Once all the differences have been added for HV2, click the 'save' button. Click the 'close' button.
- 14.4.16 A second qualified mtDNA analyst confirms that the sample is eligible for SDIS/NDIS and that the profile data has been added correctly. If so, the sample is verified by using the drop down menu to select the correct user name in the verified by column, the specimen category is changed to the correct category, the sample is marked for upload if eligible, and the specimen detail report is printed. The second qualified mtDNA analyst will use DNA QR-13A to document that the profile was entered correctly, it meets eligibility requirements, and if it has been marked for upload. The first qualified mtDNA analyst will initial and date the specimen detail report to indicate that the correct specimen category was chosen.
- 14.5 Y-STR Data Entry**
- 14.5.1 Launch Analyst Workbench and select Specimen Manager. Select 'STR/Y-STR Data Entry'.
- 14.5.2 Enter the specimen ID (case and item number). This information must be entered to continue adding additional details to the profile.
- 14.5.3 For specimen category use the drop down arrow to select the "other" category.
- 14.5.4 Verify the user name (assigned to) is correct.
- 14.5.5 No data is added to the Case ID information field.

- 14.5.6 Using the drop down arrow choose if the source is identified (Yes or No).
- 14.5.7 Using the drop down arrow choose yes or no for partial profile. (This is a required field but will default to 'no' if not chosen).
- 14.5.8 In the lower half of the screen, below the STR loci, are the loci for Y-STRs. Click in the empty field for 'Reading #1' for the first Y-STR locus. Enter in the correct allele. Press the enter key or click in the empty field for the next locus ('Reading 1'). Enter in the correct allele. Continue to do this for all of the Y-STR loci. Click in the empty field for 'Reading #2' for the first Y-STR locus. Enter the correct allele. Press the enter key. Enter in the correct allele for this next locus ('Reading 2'). Continue to do this for all of the Y-STR loci.
- 14.5.9 Alternately, after entering the allele for 'Reading 1' for the first locus, the user can hit the tab key to enter in the same allele for 'Reading 2' and then proceed to the next locus.
- 14.5.10 When done entering all the Y-STR alleles, click 'Save' and then click 'Close'.
- 14.5.11 A second qualified Y-STR analyst confirms that the sample is eligible for SDIS/NDIS and that the profile data has been added correctly. If so, the sample is verified by using the drop down menu to select the correct user name in the verified by column, the specimen category is changed to the correct category, the sample is marked for upload if eligible, and the specimen detail report is printed. The second qualified Y-STR analyst will use DNA QR-13A to document that the profile was entered correctly, it meets eligibility requirements and if it has been marked for upload. The first qualified Y-STR analyst will initial and date the specimen detail report to indicate that the correct specimen category was chosen.
- 14.6 Adding Metadata Through Specimen Manager**
- 14.6.1 Launch Analyst Workbench, click on Specimen Manager. Query the correct specimen and select it.
- 14.6.2 Click the 'Edit Specimen Metadata' button on the toolbar. Alternately, you can right click on the specimen and select 'Edit Metadata'. Note: the metadata options will be different for different specimen categories. The three specimen categories are Missing Person, Unidentified Person, and Biological Relative of Missing Person.

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- 14.6.3 The Specimen Data Summary is read-only (this information has already been entered for the profile). Select male or female. Select the Ethnic Group by using the drop down menu. **For Biological Relatives**, sex and ethnic group are the only two details that can be added for metadata under the physical characteristics tab. There is an additional tab for 'Custom Metadata' where more information can be added.
- 14.6.4 **For Unidentified Persons**, enter the age range for the unidentified remains (how old was this person at the time of their death). Enter the height in centimeters. For specimen origin, use the drop down arrow to select 'whole body', 'body part', or 'not specified'. If there are any physical anomalies and/or scars/marks/tattoos, click the appropriate square and type the information into the corresponding text box. If one or both of the squares are checked, information must be typed into the corresponding box. Add any additional information into the comments field. Click on the 'Location' tab. Click in the 'Date of Recovery' field. The date can be entered manually (mm-dd-yyyy) or the calendar function can be used (calendar will automatically open when you click in the empty field). Click in the 'Geographic Location of Recovery' field. This field is intended for GPS coordinates which can be either entered manually (note: south and west are negative values) or copied and pasted from another source. Click in the 'City' field and type in the city name. Use the drop down arrow to select the state. Click in the 'Location' field. Enter relevant information such as an address or a description of the location where the remains were found. Click 'Save'. Click 'OK'.
- 14.6.5 **For Missing Persons**, click on the 'Date of Birth' field and enter the information (mm-dd-yyyy). Enter the height in centimeters. If there are any physical anomalies and/or scars/marks/tattoos, click the appropriate square and type the information into the corresponding text box. If one or both of the squares are checked, information must be typed into the box(es). Click on the 'Location' tab. Click in the 'Date of Last Contact' field. The date can be entered manually (mm-dd-yyyy) or the calendar function can be used (calendar will automatically open when you click in the empty field). Click in the 'Geographic Location of Last Contact' field. This field is intended for GPS coordinates which can be either entered manually (note: south and west are negative values) or copied and pasted from another source. Click in the 'City' field and type in the city name. Use the drop down arrow to select the state. Click in the 'Location' field. Enter relevant information such as an address or a description of the location where the missing person was last seen. Click 'Save'. Click 'OK'.
- 14.6.6 A second qualified DNA analyst confirms that the metadata was entered correctly. The second qualified DNA analyst will use DNA QR-13A to document that the metadata was verified.

## **14.7 Pedigree Trees**

- 14.7.1 Pedigree trees can be created even if no DNA profiles have been generated yet. In the Analyst Workbench, click on Pedigree Manager. Click on the 'new' button icon (left most icon on the top toolbar). For pedigree trees with one typed node, select 'Single Typed Node' for the Pedigree Category. For pedigree trees with more than one typed node, select 'Missing Persons' for the Pedigree Category.
- 14.7.2 A single square with a question mark in it will appear in the Pedigree Tree Designer tab. This represents the missing person or unknown node (default is male). If the missing person is female, right click on the square and select 'change sex'.
- 14.7.3 To add family members to the missing person, right click on the square or circle, click 'add relationship' and then choose the type of relative you are adding to the pedigree tree. If a sibling is being added, parents will automatically be added to the pedigree tree. Continue to create family members to account for all the profiles that will eventually be added to the pedigree tree.
- 14.7.4 In the process of creating the tree, there will be nodes that will go untyped (no data associated with it). Leave these nodes as a circle or square with an 'X' in the center. When done, click the 'save' icon on the top toolbar. This will prompt a message box to name the pedigree tree. Name the tree and click 'Save'. Click 'OK'. A printed copy of the pedigree tree shall be placed in the case jacket. See section 14.10.2 for printing pedigree trees.
- 14.7.5 Note: A square represents a male. A circle represents a female. A green shape within the node represents STR data. A blue shape within the node represents Y-STR data. A red shape within the node represents mtDNA data.

## **14.8 Associating a Profile to a Node on a Pedigree Tree**

- Note: Before associating a profile to a pedigree tree node, ensure the sample has been entered correctly, verified by a second qualified DNA analyst and marked for upload. Once associated to a tree, a profile cannot be altered unless it is unassociated from the pedigree tree first.
- 14.8.1 With the pedigree tree open (or see section 14.7 to create a new tree), click on the Specimen Manager tab located next to the 'Pedigree Tree Designer' tab.

- 14.8.2 Find the correct sample, left click on the sample and drag it into the 'Pedigree Tree Designer Tab'. Once in 'Pedigree Tree Designer' and continuing to hold down the left mouse button, release the button once the mouse arrow is over the node to associate the specimen information with that node.
- 14.8.3 Click 'Save'. Name the tree (if this is a new tree). Click 'Save'. Click 'OK'.
- 14.8.4 When a specimen is first associated to a pedigree tree, a validation check is automatically performed. This check will verify the sex of the pedigree node, confirm that the specimen associated to a node is not associated to another node on the same pedigree tree, and verify that the specimen category is configured correctly. If the pedigree is not verified determine the problem and correct if data was entered incorrectly. In order to correct some pedigrees it may be necessary to contact the submitting agency to confirm the relationships of members of the pedigree.
- 14.8.5 A second qualified mtDNA analyst confirms that the pedigree tree is eligible for SDIS/NDIS and that the profile data has been associated correctly. The second qualified mtDNA analyst will use DNA QR-13A to document that the pedigree tree was created correctly and it meets eligibility requirements. If so, the pedigree tree is marked for upload and the pedigree tree report is printed. The first qualified mtDNA analyst will initial and date the pedigree tree report to verify the 'marked by' date.
- 14.9 Associating Metadata to a Node on a Pedigree Tree**
- 14.9.1 In Pedigree Manager, double click on the correct pedigree or select the pedigree from the list and click the 'open' icon on the toolbar.
- 14.9.2 With the pedigree tree open, right click on the appropriate node and choose 'edit node Metadata'.
- 14.9.3 The Pedigree Metadata Summary is read only. The metadata 'sex' option is determined by the sex of the pedigree tree node and is also read-only. Click on the drop down arrow and choose the ethnic group.
- 14.9.4 Refer to section 14.6.5 for Missing Persons. Refer to section 14.6.4 for Unidentified Persons. Refer to section 14.6.3 for Biological Relatives.
- 14.10 Checking and Managing Pedigree Trees**

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- 14.10.1 Consistency checks: Consistency checks will verify that the DNA data is consistent between relatives within the pedigree tree. A consistency check can be done manually by clicking the red arrow icon on the toolbar while in Pedigree Manager and with a pedigree tree open. A window will appear if the tree is valid with no inconsistencies. Click 'OK'. A consistency check is automatically done when a pedigree is saved. If there are any inconsistencies, the pedigree cannot be saved until the proper corrections are made.
- 14.10.2 Exporting and printing a pedigree tree: A Pedigree Detail Report will contain information about the pedigree and a graphical display of the pedigree tree. To print the report: in pedigree manager select the pedigree, either right click on the selected pedigree and select print pedigree or select the print icon on the tool bar.
- 14.10.3 Renaming a pedigree tree: In Pedigree Manager, right click the pedigree tree to be renamed and select 'Rename Pedigree'. Enter the new name for the pedigree and click 'Save'. Click 'OK'.
- 14.10.4 Deleting a pedigree tree node: With the pedigree tree open (in Pedigree Manager) right click on the node being deleted and select 'Delete Node'. Click 'Yes'. If this node is crucial to the integrity of the pedigree tree, an orphaned node will be seen within the pedigree screen and the tree cannot be saved.
- 14.10.5 Deleting a pedigree tree: In Pedigree Manager, select the pedigree tree from the list. Click the 'delete' icon (red X) on the toolbar. A Pedigree Delete Report will be generated and sent to the CODIS Message Center.
- 14.10.6 Following all changes to pedigree trees a new copy of the tree is printed and placed in the case jacket.
- 14.11 Searching at SDIS and NDIS**
- 14.11.1 When DNA records are uploaded into an NDIS Missing Person Index, new or modified DNA records are marked to indicate that they have not been searched. On a schedule determined by the NDIS Custodian, an Autosearch will be run to search previously unsearched DNA records.



- 14.11.2 Only complete profiles (see section 14.3) will be autosearched. Additional DNA technologies (such as mtDNA and YSTR) should always be considered for missing persons, relatives of missing persons and unidentified human remains as appropriate. However, the inclusion of DNA data from an additional relevant technology is required for the searching of a single family reference sample at NDIS. For example, if the missing person is a female, then YSTR technology would not be relevant. Use of an additional relevant technology will also ensure the most robust search possible.
- 14.11.3 SDIS shall be configured to search the following Indexes against each other:

**SDIS and NDIS Searchable Indexes**

Index	Forensic and Forensic Mixture	Convicted Offender, Arrestee, Detainee and Legal	Unidentified Human (Remains)	Missing Person	Relatives of Missing Person and Pedigree Tree
Forensic and Forensic Mixture	X	X	X	X	
Convicted Offender, Arrestee, Detainee and Legal	X		X	X	
Missing Person	X	X	X		X*
Relatives of Missing Person* and Pedigree Tree			X	X*	
Unidentified Human (Remains)	X	X	X	X	X

\* State searchable index only

- 14.11.4 In a Pedigree Tree Autosearch, unidentified human remains (UHR) are compared to Pedigree Trees to determine the statistical likelihood that the unidentified human remains are those of the relative represented by the missing person node of the tree. In addition to the Joint Pedigree Likelihood Ratio (JPLR) produced by STR results, additional technologies (Y-STR and/or mtDNA) may each provide a likelihood ratio that contributes to the overall Combined Probability Likelihood Ratio (CPLR). For this search, the NDIS Custodian establishes a statistical threshold in order to limit the search results to only the most likely UHR-Pedigree Tree pairings. SDIS and NDIS thresholds are 500 for CPLR, 1 for JPLR, 1 for mtDNA LR, and 1 for Y-STR LR. In the event that Likelihood Ratios need to be calculated, a qualified individual will be contacted.

- 14.11.5 The laboratory responsible for a Pedigree Tree shall receive a rank message that includes all UHRs exceeding the statistical threshold. Conversely, the laboratories responsible for the UHR will receive a rank message including Pedigree Trees to which the UHRs likelihood ratio exceeded the threshold. The laboratory responsible for the UHR will not receive messages containing UHRs from other laboratories which may be more strongly associated to the Pedigree Tree.

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