DNA Type Lookup Tool Instructions Updated: 9/27/05

Allele codes are a short hand alphabetic notation used by the NMDP to capture DNA based allele combinations in our database. Once a new allele combination is encountered, an allele code is assigned to that allele combination and it's then made available (activated) for use at that specific locus, and for that specific allele family. The following guidelines will assist in the use of the DNA Type Lookup Tool and present some of the unique features available. This tool accesses data from allele code tables which are refreshed on a daily basis. Requests for new allele codes, or activation of an existing code at a different locus, can be submitted using the email link that appears at the bottom of the DNA Type Lookup Tool page. However, for requests of five or more allele codes or activations, you may want to submit them all in one email message or as an Excel attachment to <u>new-allelecodes@nmdp.org</u>. A glossary of terms and definitions has been provided at the end of this instruction document.

What can I use this tool for?

- 1. To expand an allele code to see which alleles it contains.
- 2. <u>To find a code for a combination of alleles.</u>
- 3. <u>To see whether an allele code has been activated for use with a specific locus and allele family.</u>
- 4. To view all the alleles in a particular allele family.
- 5. To view all possible loci for which a code has been activated.
- 6. <u>To request the creation or activation of an allele code by the NMDP.</u>

The <u>Features</u> section below contains additional information about this tool.

QUESTION 1: How do I expand an allele code to see the alleles it contains?

Option one:

- 1. Select the locus of interest.
 - 1.1. Click on B
- Enter the allele family followed by the allele code in the data entry box.
 2.1. Enter 27EKN for example.
 - 2.1. Enter 2/EKN for example.
 - 2.1.1. **NOTE:** Allele codes can be entered using either capital or lowercase letters. All capital letters should be used for reporting final results containing allele codes.
- 3. Click the submit button.
 - 3.1. The allele code will be presented and all the alleles it contains will be listed.
- 4. Expanded allele lists may appear with either two or four digits. Two digits are used for <u>generic</u> codes and four digits are used for <u>allele specific</u> codes (allele combinations which contain a null allele, for example).

Option two:

- 1. Enter just the letter portion of the allele code in the data entry box. 1.1. EKN, for example.
- 2. Click the submit button.
 - 2.1. The allele code will be expanded to display all the alleles it contains and a list of all loci and allele families where that combination has been <u>activated</u> for use.

QUESTION 2: How do I find a code for a combination of alleles that I want to report?

- 1. Select the locus of interest (A for example)
- 2. Enter into the data entry box the first full allele of the allele combination, followed by the list of alleles, using the last two digits only (three in case of alleles with an expression character, null [N], low expression [L], and soluble [S]) (i.e. 1101/02/03)
 - 2.1. **NOTE:** Entering all four digits for the first allele, designates the <u>allele family</u> of interest for that locus (e.g. entering 2402/04 returns A*24BD)
 - 2.2. **NOTE:** If an allele combination contains alleles from two separate families, the first allele from the second family must contain all four digits (e.g. 0101/02/04/0202/05-07)
 - 2.3. **NOTE:** Alleles may contain more than five or more digits (e.g. 13012). In these cases do not enter any digits after the fourth number unless it is an expression character such as N, L, or S (N=null, L=low expression, S=soluble) (e.g. 4423N).
 - 2.4. **NOTE:** When entering allele combinations to search for a code, enter ALL the alleles in the combination, be sure to include expression characters in the alleles. Shortening the list will result in a code which does not represent the combination obtained by laboratory analysis. Using a code which has the "Best Fit" to you allele combination is prohibited for the same reason.
 - 2.5. Entering just the last two digits of an allele list will result in an allele code and all families within that locus selected for which that code has been activated. (e.g. Entering 01/02/03 returns MN and a list of families in the A locus where MN applies will appear below). See several <u>data entry formats</u> in the Features section below.
- 3. Hit the submit button.
 - 3.1. If an allele code exists, it will be provided followed by the list of alleles it contains and whether or not the code has been <u>activated</u>.
 - 3.2. **NOTE:** Codes listed as **NOT ACTIVATED**, will not be recognized by the STAR system and must be activated by the NMDP before they can be used.
- 4. If no code exists, or a code is not activated, see Question 6.

QUESTION 3: How can I tell if an allele code has been activated for use with a particular locus and allele family?

- 1. Select the locus of interest.
- 2. Enter the allele family followed by the allele code in the data entry box (i.e. 24BD).
- 3. Click the submit button.
 - 3.1. The allele code will be expanded and **ACTIVATED** or **NOT ACTIVATED** will appear below the text.
- 4. To activate a code, see Question 6.

QUESTION 4: How do I view all the alleles in a particular allele family?

- 1. Select the locus of interest.
- 2. Enter the allele family followed by XX.
 - 2.1. A*01XX, B*07XX, DRB1*13XX, etc.
 - 2.2. XX is a special allele code assigned to represent all of the alleles currently active in our system (at the time of your request), for a particular allele family and locus.
- 3. Click the submit button.
 - 3.1. A list of all the alleles for that locus and allele family will appear.

QUESTION 5: How do I view all possible loci for which a code has been activated?

- 1. Select "None" for the locus.
- 2. Enter just the letter portion of a code, "MN" for example.
- 3. Click the submit button.
 - 3.1. A list will appear of all the loci and allele families for which MN has been activated.

QUESTION 6: How do I request the creation or activation of an allele code?

- 1. **NOTE:** When entering allele combinations to search for a code, enter ALL the alleles in the combination, be sure to include expression characters in the alleles. Shortening the list will result in a code which does not represent the combination obtained by laboratory analysis. Using a code which has the "Best Fit" to you allele combination is prohibited for the same reason.
- 2. If an allele combination is unrecognized by the tool, the message "No code exists for the above combination!" will appear. A link will appear below this message for you to electronically submit the combination to the NMDP for allele code creation.
 - 2.1. Clicking the link will open a new message window of your default email application. In the body of the message, please provide your name, phone and laboratory or center name. The allele combination entered into the tool is captured and inserted into the body of the message.
 - 2.2. Click send; your request is submitted to the department of Scientific Services at the NMDP for review.
- 3. If a code is listed as **NOT ACTIVATED**, a link will appear below for you to electronically submit the combination to the NMDP for review and activation.
 - 3.1. Clicking the link will open a new message window of your default email application. In the body of the message, please provide your name, phone and laboratory or center name. The allele combination entered into the tool is captured and inserted into the body of the message.
 - 3.2. Click send. Your request is submitted to the Scientific Services department at the NMDP for review.

FEATURES:

- 1. <u>Reset button</u> This clears all fields.
- 2. <u>Hint links</u> Hints appear below the tool that provide examples of how to enter data. 2.1. A new hint appears each time the reset button is pressed.
- 3. <u>Warning messages</u> A warning will appear if you have entered an invalid allele.
 - 3.1. This may indicate that the allele entered is either a "null" allele, a new allele not yet entered into our system, or an allele that has been deleted from our system.
- 4. <u>Activation status</u> When a locus is selected and an allele combination entered, the tool will return a code and whether or not the code has been "<u>activated</u>" for that specific locus and allele family.
 - 4.1. As codes are created, they are made available (or activated) for use with a specific allele combination, and at a specific locus.
 - 4.2. A code may exist for a particular allele combination, and still not be activated for the locus you are searching.
 - 4.3. The new tool will display "ACTIVATED" and "NOT ACTIVATED" accordingly.
- 5. <u>Data entry formats</u> Allele code strings may be entered in several different formats which appear below:
 - 5.1. Allele combinations can be separated with a plus, slash, comma or space. Examples of each are as follows:
 - 5.1.1. 0101+0102+0103
 - 5.1.2. 0101,0102,0103
 - 5.1.3. 0101/0102/0103
 - 5.1.4. 0101 0102 0103
 - 5.2. After the first complete allele, for family designation, the remaining alleles may be entered using only the last two digits.
 - 5.2.1. 0101/02/03
 - 5.2.2. 0101,02,03
 - 5.3. If no locus is specified, the tool will return the code for the combination entered and list all possible loci where that allele code is active.
 - 5.4. Long , consecutive allele combinations may be entered using a dash and the tool will remove all deleted alleles and correctly label all alleles which contain an expression character.
 - 5.4.1. 0201-34 will return all the alleles from 0201 through 0234
 - 5.5. Combinations of allele codes may be entered.
 - 5.5.1. 02GSA+14-24 will return the alleles in the GSA code plus all the alleles from 0214 through 0224.
 - 5.5.2. 02AB+CD will return the alleles in the AB code plus those in CD.
 - 5.5.3. 02CWN+0224 will return the alleles in the CWN code and add 0224.

GLOSSARY:

<u>Allele Code</u> - A shorthand alphabetic notation used to represent a combination of alleles. B*27EKN. EKN is the allele code for 05/13.

<u>Allele Family</u> - Genetic groupings within a single locus. All families are assigned a numeric identifier. B*27EKN - 27 is an allele family within the B locus.

<u>Locus</u> - position of a gene on a chromosome. B*27EKN - B*locus is the genetic region of interest.

<u>Generic</u> - An allele combination which may be applied to more than one locus and family (e.g. MN = 01/02/03).

<u>Allele Specific</u> - An allele combination which contains a null allele or one which has alleles from two or more allele families (e.g. 04KBG = 0401/0409N, 02TKK = 0202/0301).

<u>Activated</u> - Is a status indicating that a code is available for use with a specific locus and with a specific allele family.

<u>Not Activated</u> - Is a status indicating that a code is not available for use with the specific locus and with the specific allele family requested.