## Help File for CANDELA Face Phenotyping program

Version 1.4, created 19 May 2016

## Important usage information:

This software was developed at University College London for use in the CANDELA project: <a href="http://www.ucl.ac.uk/silva/candela">http://www.ucl.ac.uk/silva/candela</a>

Software developed by Kaustubh Adhikari. Drawings by Macarena Fuentes-Guajardo and Elizabeth Guajardo Celsi.

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If using the software, please cite:

Adhikari, K. et al. A genome-wide association scan implicates DCHS2, RUNX2, GLI3, PAX1 and EDAR in human facial variation. Nat. Commun. 7:11616 doi: 10.1038/ncomms11616 (2016).

Phenotypes and phenotype categories are derived from Ritz-Timme, S. et al. A new atlas for the evaluation of facial features: advantages, limits, and applicability. Int J Legal Med 125, 301-6 (2011).

For bug reports, or recommendations for improvement, please contact <u>k.adhikari@ucl.ac.uk</u>. In case of bugs, please also attach the 'worklist.mat' file and MATLAB error messages.

## Steps for running the program:

1. You need to have MATLAB installed to be able to run this program. It has been tested to run on MATLAB versions R2010b, R2013b, R2014b, and R2016a, each showing slight variations in the user interface appearance.

If you don't have MATLAB installed, you need to install the freely available MATLAB Compiler Runtime (MCR) and ask the software author to provide a compiled version of the software.

- 2. A screen with resolution at least 1200×800 is necessary to run this program.
- 3. This GUI (graphical user interface) program has two components:

face\_phenotyping.fig and face\_phenotyping.m.

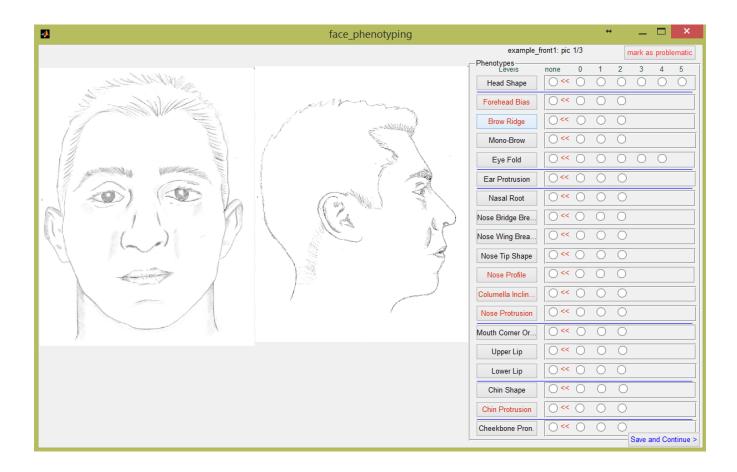
The accompanying file help mat contains sample images for each phenotype.

- 4. Create a folder in which to keep the program and all the photos. Here, it will be called 'main'.
- 5. Copy the face\_phenotyping.fig and face\_phenotyping.m files to the 'main' folder. Also copy the file help.mat to this folder.
- 6. Create two sub-folders within this parent folder. They need to be named 'front' and 'side'.
- 7. For every person that you want to phenotype, copy the frontal facial photograph to the 'front' folder and side facial photograph to the 'side' folder.

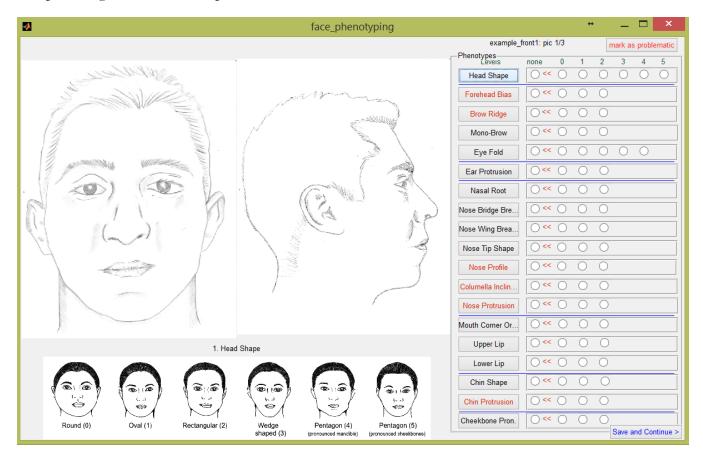
There is no restriction of file naming convention, however it is important that the files are named in such a way that the ordering of individuals is the same in the photos inside front and side folders.

Three sets of facial drawings are provided with this software as examples. The 'front' folder has files named example\_front1.png, example\_front2.png, example\_front3.png, while the 'side' folder has corresponding files named example\_side1.png, example\_side2.png, example\_side3.png.

- 8. There is no restriction on image file formats png, jpg, gif, tif, bmp etc. will all work. However it is preferable to consistently use a single file format, to avoid any mistakes in automatic file ordering.
- 9. Run the face\_phenotyping function in a MATLAB command window (without any arguments). If using a pre-compiled version, double-click face\_phenotyping.exe to run the program. The main program window will appear.
- 10. The front and side photos for the first person in the folder will be shown in the program window. Note that, at the top right of this window, it is showing 'example\_front1: pic 1/3'. It is showing the frontal file name for this person, and that it is the first picture among all the three pictures that we have in the folder.

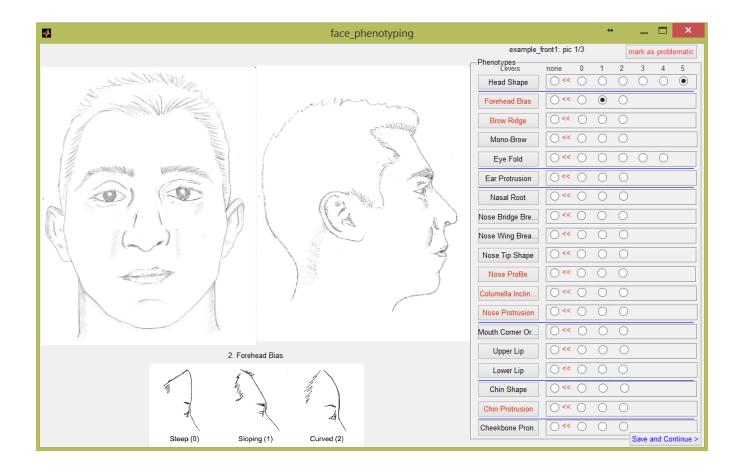


11. This program can rate 19 specific phenotypes of the face, which are listed in the window. All the phenotypes are ordinal (categorical), and most of them have three categories: 0 to 2. One phenotype has five categories and one has six. For each phenotype, you can click on the name to bring up a sample image at the bottom of the window. The sample image shows all the categories for this phenotype, their names and illustrations. In the example below, the sample image for Head Shape is shown.



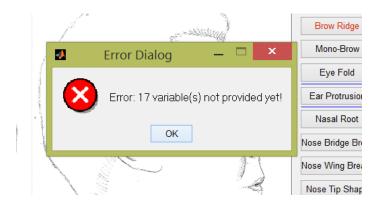
The phenotypes are grouped by the blue separator lines. E.g. the first group (with only one phenotype) corresponds to the head, the second to forehead, the third to eye & eyebrow, the fourth to ear, the fifth to nose, the sixth to lips, etc.

12. The phenotype names that are in black correspond to the frontal photo, e.g. eye fold, lip thickness, chin shape etc. The phenotype names that are in brown correspond to the side photo, e.g. forehead slope, nose profile etc. This will also be clear by looking at the sample image. In the example below, the sample image for Forehead Bias is shown.



- 13. For each phenotype, the buttons on the right corresponding to its categories. You have to click on one of the buttons to provide a rating. As an example, see the above picture, where Head Shape and Forehead Bias have been rated. You can change the ratings by simply selecting a different button.
- 14. It might happen that a particular phenotype is difficult to rate. E.g. if the ear is covered by hair, Ear Protrusion might not be visible. In that case, instead of selecting one of those categories, select the 'none' category.
- 15. If the person is difficult to rate overall, e.g. if the picture is heavily blurred, then click on the 'mark as problematic' button on top-right. This picture will be skipped, this information will be marked in the data file, and the next pair of pictures will come up in the image window.

- 16. Once ratings have been provided for all phenotypes, click on the 'Save and continue' button. The ratings for this image will be saved, and pictures for the next person will come up in the program window.
- 17. If you have not provided ratings for some phenotypes but still try to save, an error dialog will come up, mentioning how many phenotypes are yet to be rated.



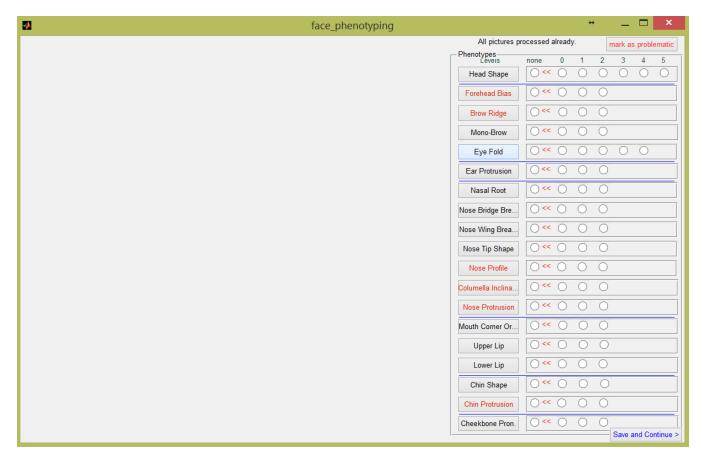
18. In the 'main' folder, two new files will be created. One is the 'worklist.mat' file, in which the program stores some information (so please do not modify or delete it). The other one is the 'data\_face.xlsx' file, which stores the ratings provided for each person. The top row contains the column names. The first column contains the codes for each person rated so far. Columns 2 to 20 contain the ratings provided for the nineteen phenotypes. The last two columns contains the time taken, in seconds, to rate this person, and the date on which this person was rated.

The numbers should range between 0 to 2 when the phenotype has 3 categories (or 0 to 5 if the phenotype has 6 categories, for example). If 'none' was selected for a particular phenotype, it will be marked as -1. If no ratings were provided for a person entirely, by using 'mark as problematic', then all the numbers will be -100.

19. While providing ratings for a person, you can close the program by simply clicking the close (X) button on the top corner. When you start the program next time, the program will take up the same person again.

E.g. if you have finished rating person 2, click on 'Save and continue' to save the ratings for this person. Person 3 will come up. You can close the program now. When you open the program next time, person 3 will come up again, and you can start providing ratings.

20. Once you have finished providing ratings for all the people, the program will tell you that all the photos are finished. If you close the program and open it some other time, you will get the same message.



21. To phenotype more people, simple copy more photographs into the 'front' and 'side' folders. E.g. if you copy 100 more images, when you run the program next time you will see the count in the header changed to '4/103'.

You can delete the older photos, but make sure that you keep a copy of the 'data\_face.xlsx' file in that case. Otherwise, when you run the program next time, the entries for all the deleted photos will be removed from the data file, so their information will be lost.

It is recommended that you back-up the 'data\_face.xlsx' file every time after using the program.

## 22. There is no 'undo' option in the program:

While rating a person, you can change your ratings by simply clicking on a different category button.

After you have saved your ratings for a person, you cannot modify the ratings. Changing it in the 'data.xlsx' file will not work, as the program rewrites the file every time.