

Introduction and Demographic Questions

Thank you for participating in this study – we are truly grateful. This project involves using “artificial intelligence” to see whether we can help build useful tools that may be relevant for different genetic conditions.

We will first ask a few general questions including whether you are willing to participate and your area of expertise. We anticipate that participation will take most people less than 15 minutes.

Next, we will show you facial images and ask you to classify whether the patient has the stated genetic condition or syndrome. The original images come from a variety of sources including medical journals and informal photographs. All images have been modified from their original format using artificial intelligence tools. Some of the images may be difficult to identify, while others may be easier. This can help us understand which types of images are easier or harder to classify.

You may take this survey on your smart device or your desktop/laptop computer.

Please do not use the internet or textbooks. We are interested in your instinctive response to each question. Once you answer a question, you will not be able to go back.

No identifiers will be collected as part of the survey. All individual results will be maintained in an anonymous format. You will not receive your individual results. We plan to describe/publish only deidentified results.

Your participation is voluntary and will not be compensated. We have been granted exemption from IRB review by the National Institutes of Health (NIH). By continuing with the survey, you agree to take part.

If you have questions, encounter any difficulties or change your mind about participation, please contact Rebekah Waikel at rebekah.waikel@nih.gov or 301.435.6558.

I am willing to participate in this study.

- I agree.

I am

- a board-certified or board-eligible Clinical Geneticist physician.
- a clinical genetics resident or fellow.

For how long (since your last residency or fellowship) have you been practicing?

- < 1 year
- 1 to 5 years
- 5 to 10 years
- > 10 years

When first encountering a patient with possible genetic condition (prior to sending genetic testing, if available), in general, how much weight do you place upon facial features in the consideration of a possible diagnosis?

- Major factor in diagnosis
- Intermediate factor in diagnosis
- Minor factor in diagnosis
- Not a factor in diagnosis

Directions

For the following questions you will view facial images of individuals who may have a genetic syndrome. We will ask about 4 genetic syndromes: Angelman syndrome, Noonan syndrome, Williams syndrome, and 22q11.2 deletion syndrome. Below each image you will select either one of these 4 syndromes or unaffected. After you have selected your answer, click the arrow at the bottom right of the window to enter your answer and progress to the next question.

Facial Images



Select whether the image shows a person with one of the syndromes or is unaffected.

- 22q11.2 deletion syndrome Angelman syndrome Noonan syndrome
- Williams syndrome Unaffected



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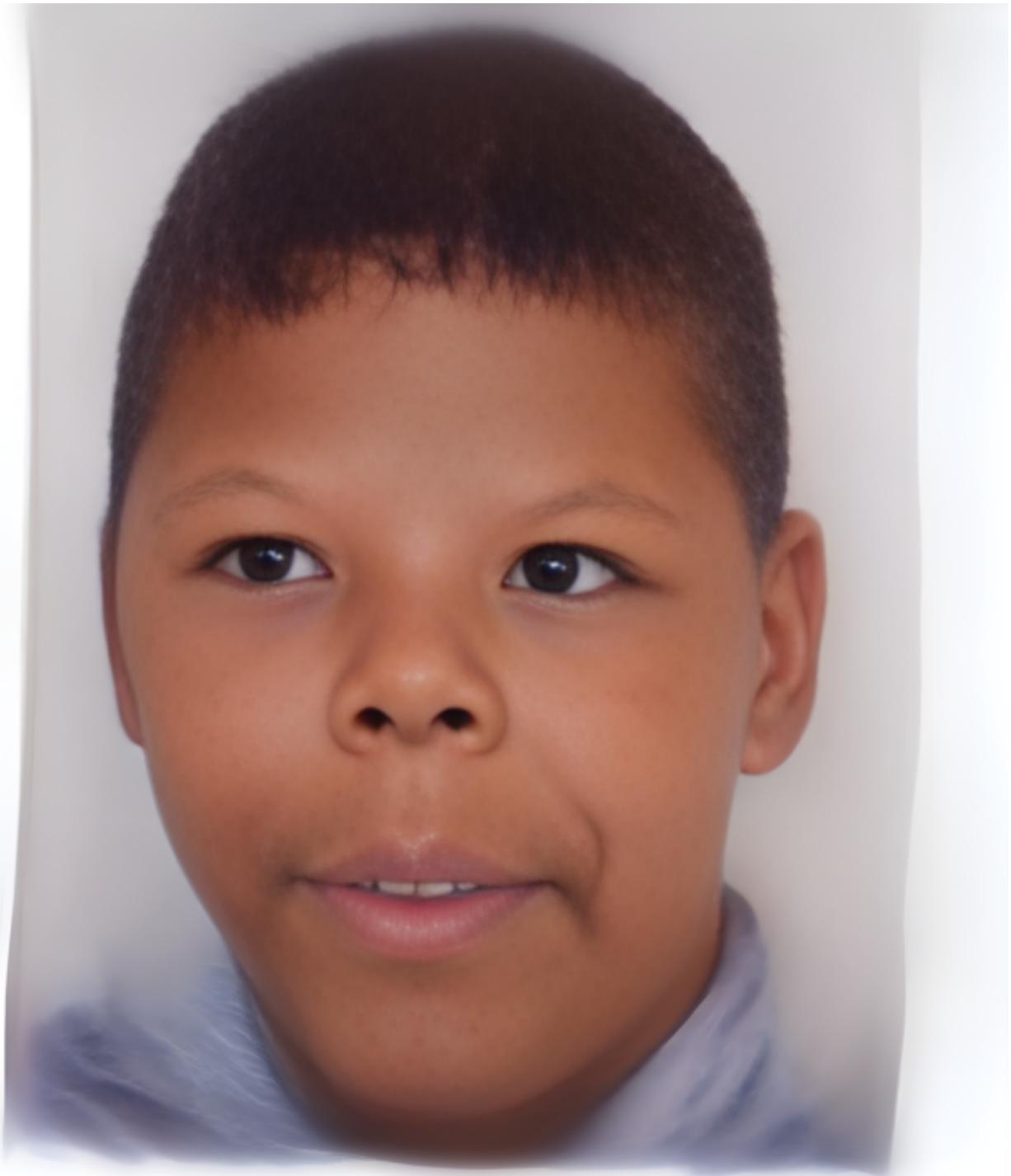
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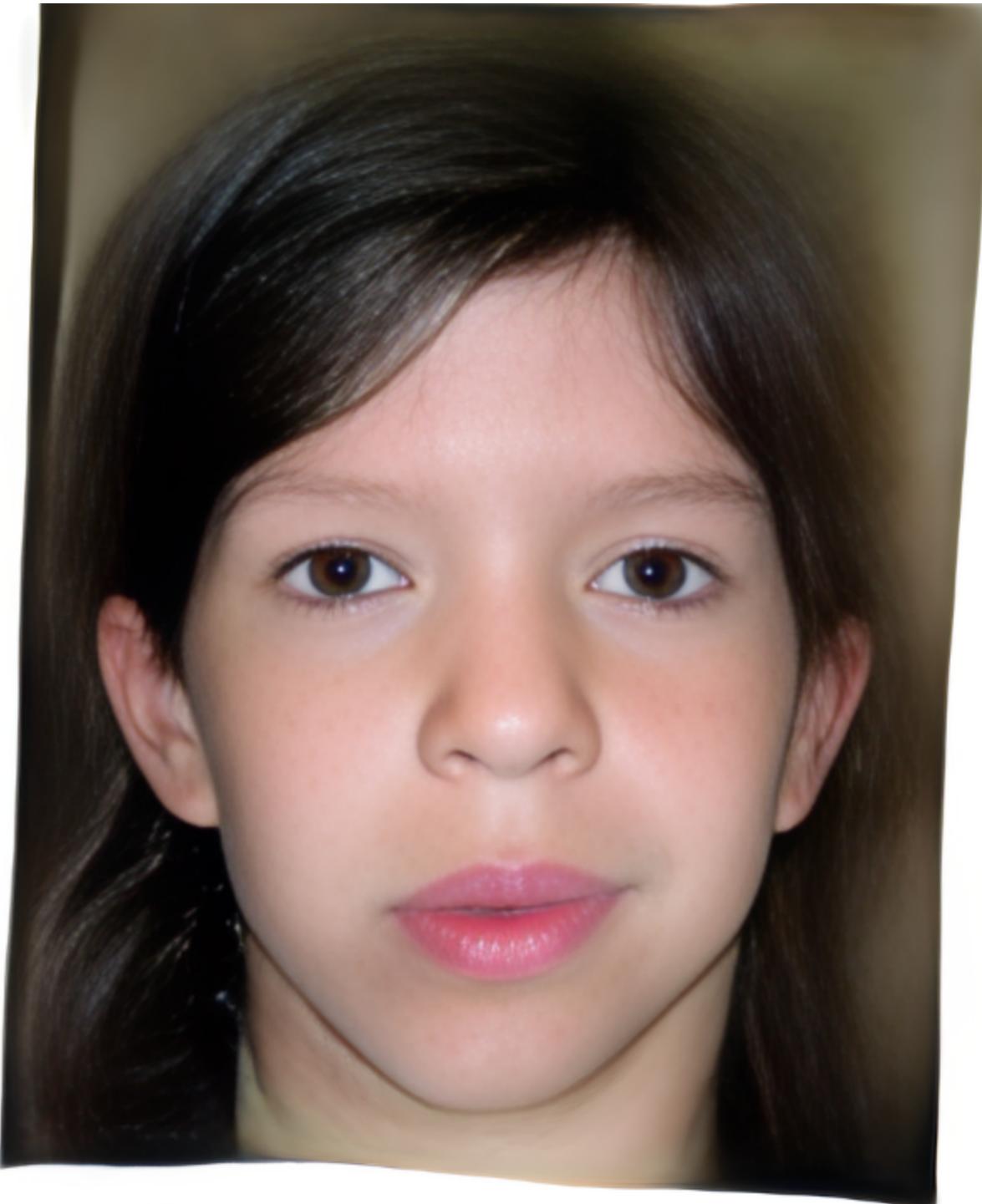
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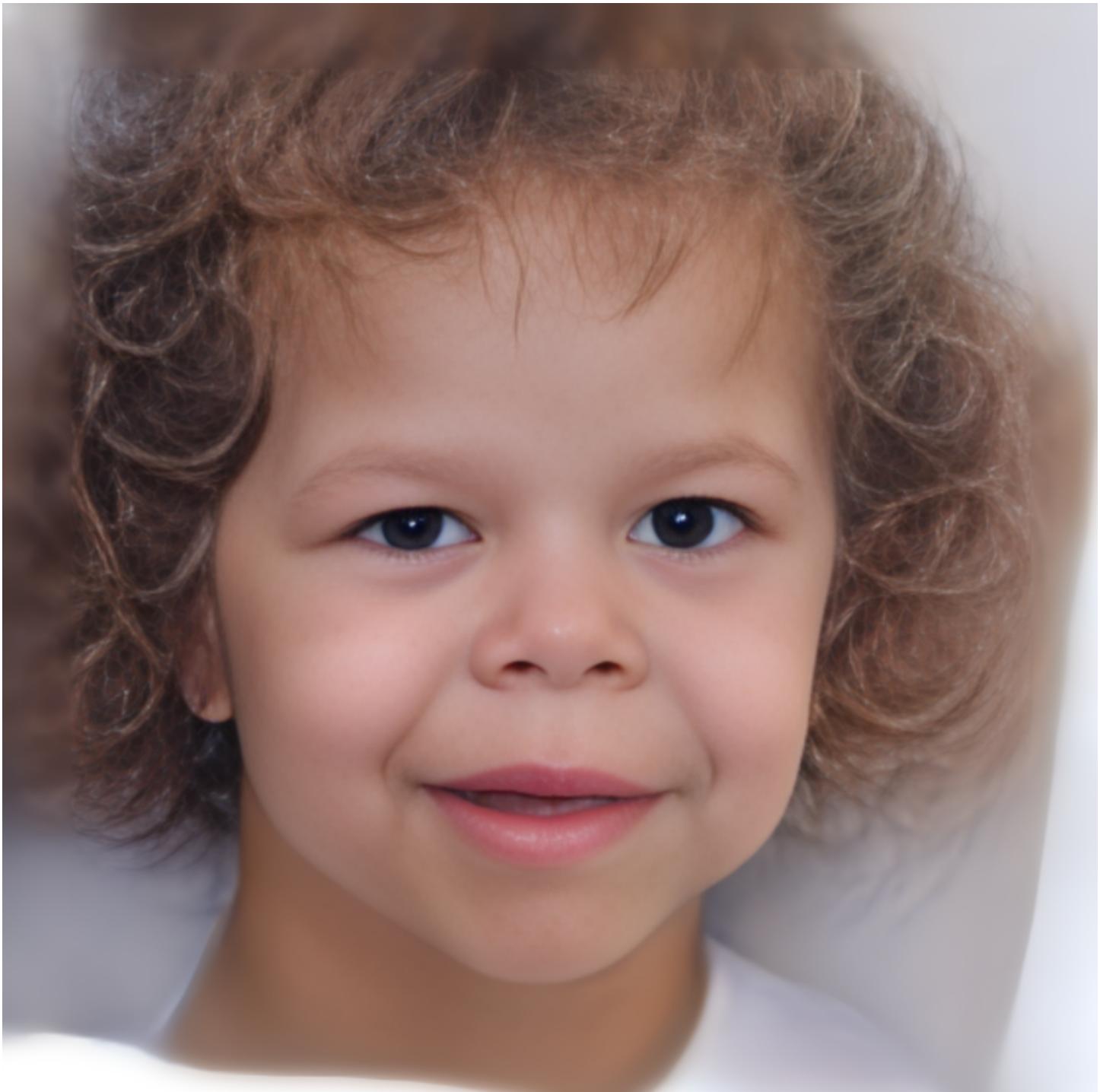
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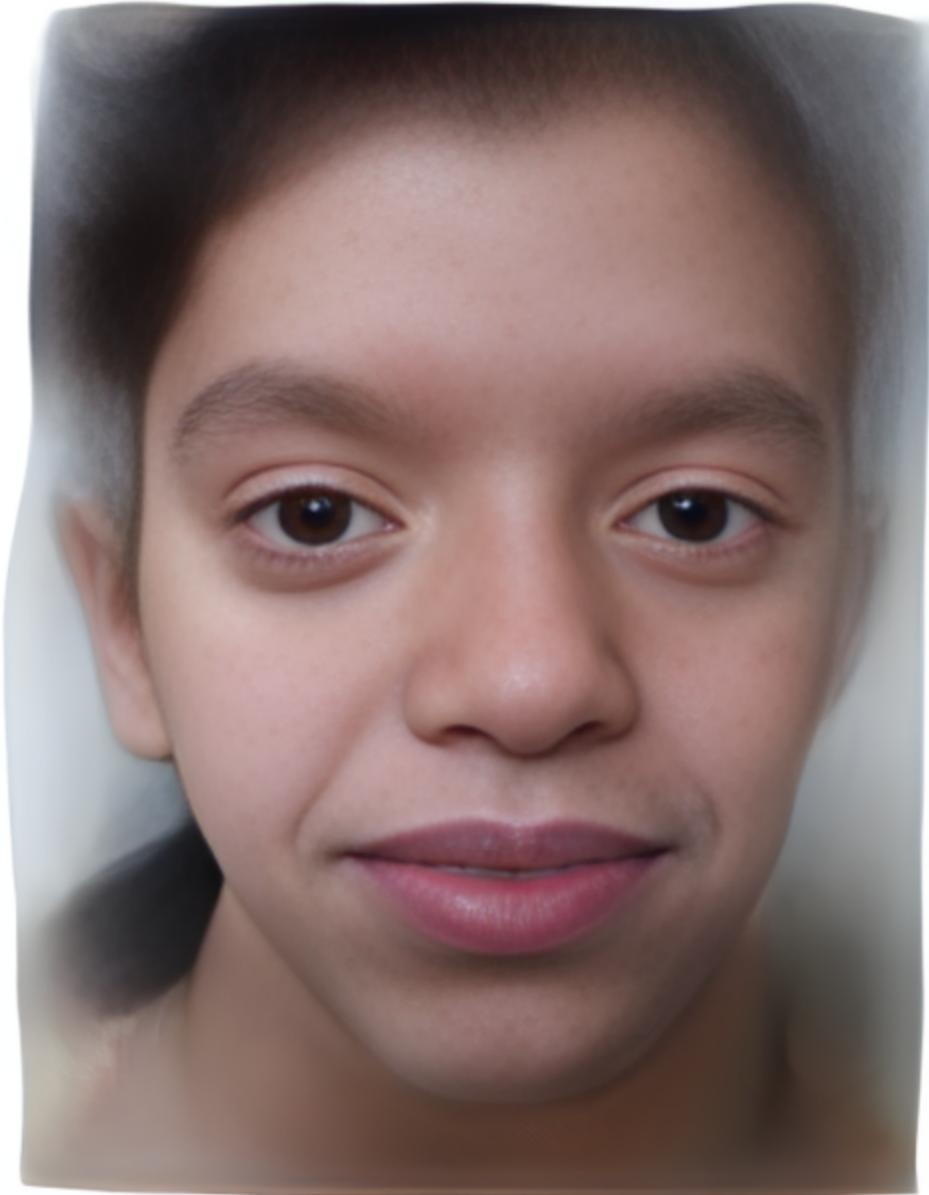
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Thank you

End of survey. Thank you.

