

Consenting language

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, your background, and your opinion about AI tools and facial diagnostics. We will show output of a facial phenotyping tool on a series of images and ask you to “classify” or diagnose a facial image of a patient as having one of five genetic conditions or unaffected. We will show you examples in which the artificial intelligence tool correctly identified and incorrectly identified the images.

We anticipate that participation will take most people less than 20 minutes.

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.

I agree to participate

- Yes
- No

Demographic Questions from AI attitudes

I am

- a board certified or board eligible medical professional (MD, DO, GC, NP, or PA)
- a genetics trainee (resident or fellow)
- medical student
- other

Please indicate your profession

Please select your medical profession (most specialized certification).

- Medical Geneticist
- Physician
- Genetic Counselor
- Physician Assistant
- Nurse Practitioner
- Other

Please write in your medical specialty (e.g., pediatrician, endocrinologist, psychiatrist).

Please write in your medical profession.

For how long (since you became board eligible) have you been practicing?

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

How would you describe your institution?

- Community based hospital
- Academic medical or research center

- Molecular diagnostic company
- Telehealth company
- Other

Please describe your institution.

What year are you in your genetics training program?

- Year 1
- Year 2
- Year 3
- Year 4 or more

What year are you in your medical school program?

- Year 1
- Year 2
- Year 3
- Year 4

How often do you use AI tools (e.g., Face2Gene) in the diagnosis of patients?

- Routinely
- Sometimes
- Have tried once or twice
- Never
- Unsure

In your opinion, how accurate are AI tools in the diagnosis of patients with genetic conditions?

- Highly accurate
- Mostly accurate
- Inaccurate

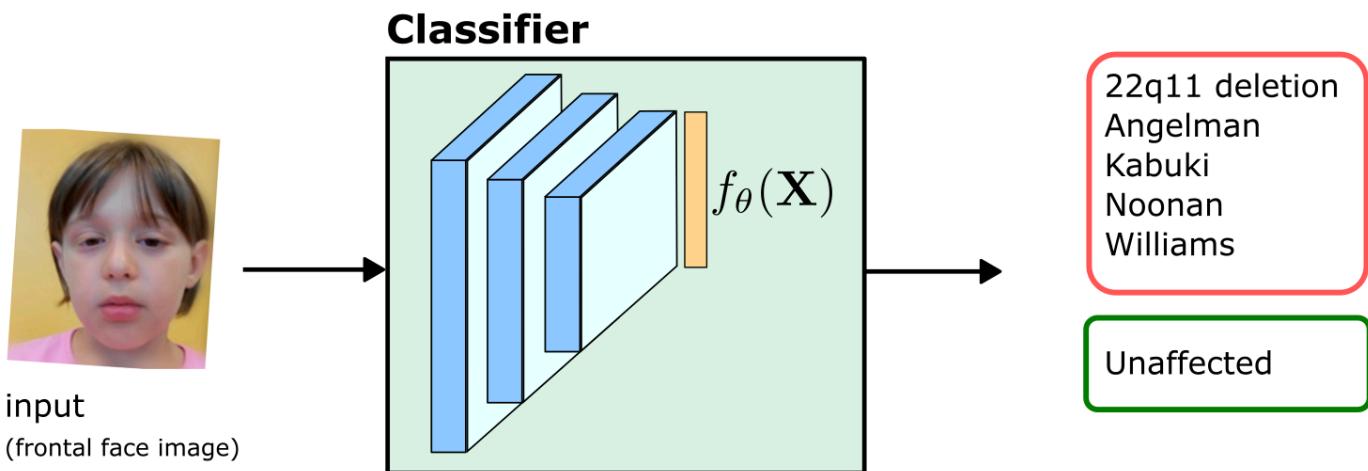
Unsure

How much weight do you place upon facial features in the diagnosis of a genetic condition?

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

Introduction to Study

We developed an AI-based tool that can analyze an image of a person and help determine whether that patient may have a genetic condition. For this study, our tool is examining five different genetic syndromes.



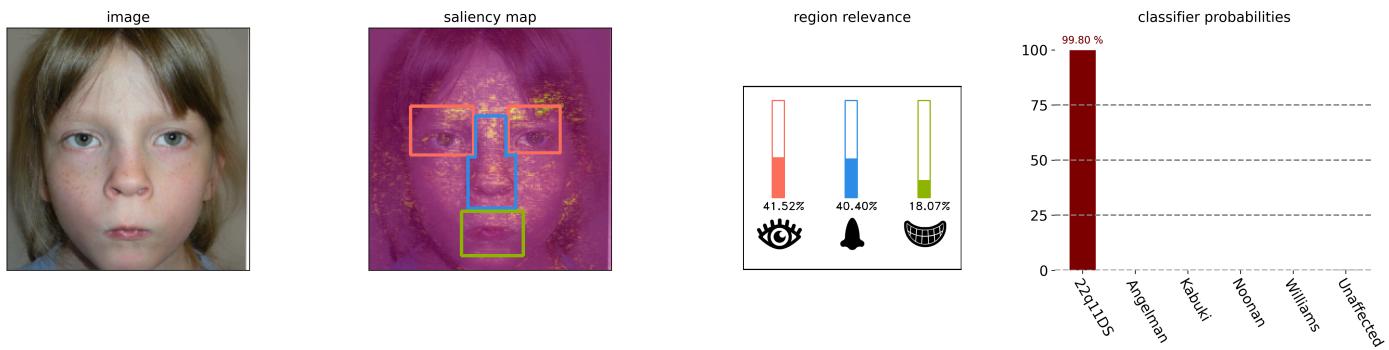
During this study, you will see 18 images of individuals who are either unaffected or who have one of the following genetic conditions:

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

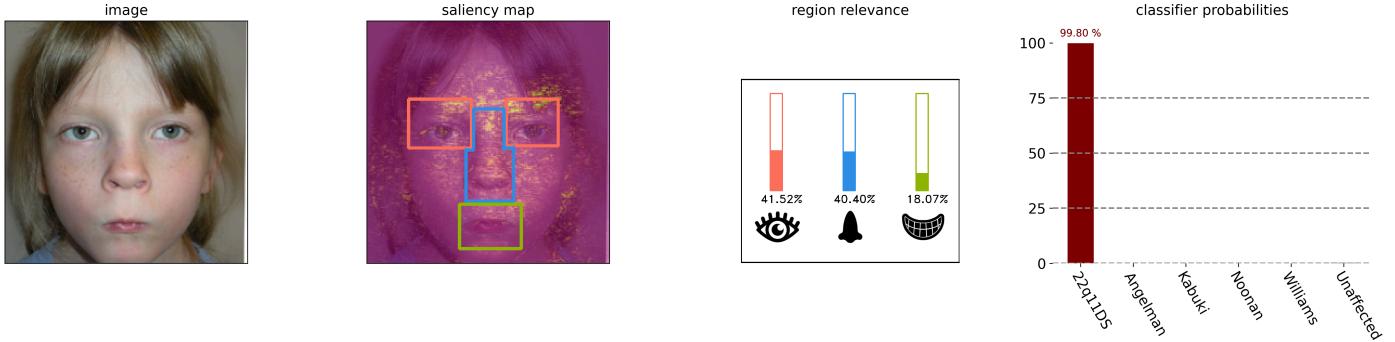
After you see each individual facial image, you will see an explanation of how the AI reached a diagnosis in term of how different face regions (eyes, nose, and mouth) contribute to the diagnosis. These are the regions that usually contain the dysmorphic features associated with these diseases. A bar graph of the prediction probability of each syndrome or unaffected is also shown.

Now let's have a look at the AI tool outputs that you will see for each image.

- Saliency Map. The saliency map (in the middle) shows the contribution of each pixel on the image to the AI model's diagnosis. The pixel color ranges from magenta to yellow (most important). Three rectangles on the image indicate the mouth, nose, and eye regions.
- Region Relevance. The region relevance displays the contribution of each region to the diagnosis. The fuller the bar, the greater the contribution.
- Probability Output. Bar graph showing the prediction probability of each of the 5 genetic conditions or unaffected.



Your task is to determine if the image represents a person with a genetic condition and select condition or unaffected followed by rate your confidence in your answer (example below). First you will answer these questions based on the image alone and then you will do this again with both the images and AI tool outputs. You will also be asked to rate the helpfulness of the outputs.



Select the name of the genetic condition.

22q11.2 deletion syndrome

Rate your confidence in the answer above.

-2 -1 0 +1 +2

Genetic Conditions

During this survey you will see images of unaffected individuals and individuals with 1 of 5 different genetic conditions.



22q11.2 deletion syndrome

Facial characteristics of 22q11.2 deletion syndrome can include: malar flattening, hooded eyelids (dermatochalasis), broad nasal bridge, ear anomalies including overfolded or squared helices, as well as other findings.



Angelman syndrome

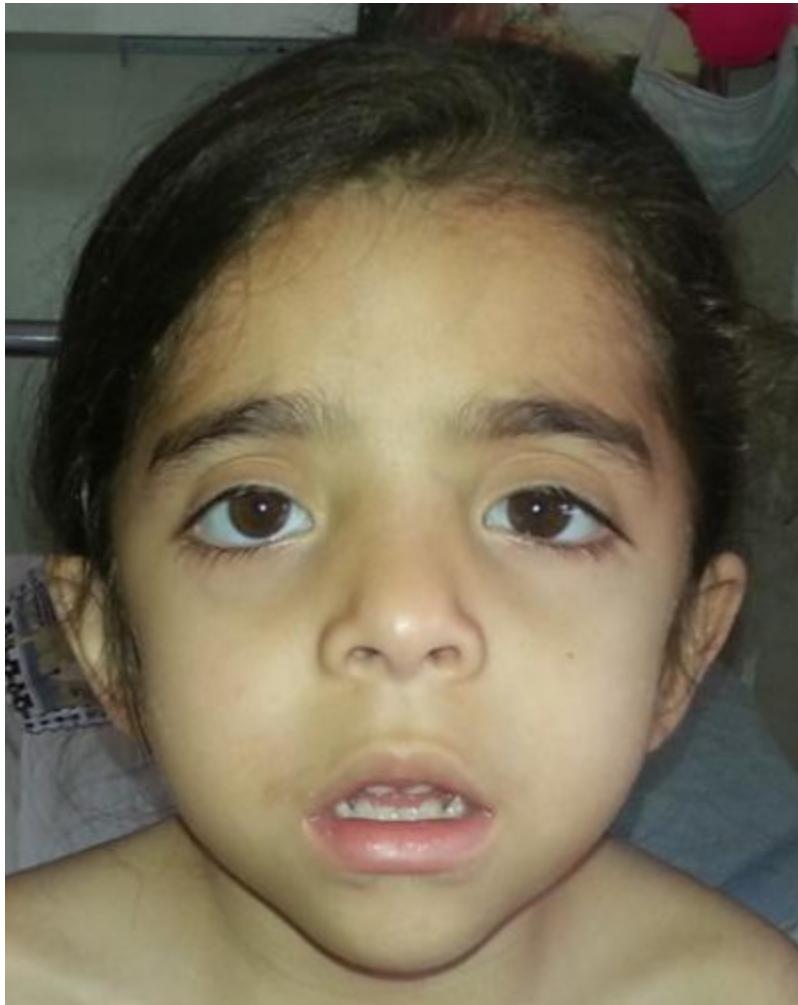
Facial characteristics of Angelman syndrome can include: deeply set eyes, pointed chin, wide mouth, as well as other findings.



Kabuki syndrome

Facial characteristics of Kabuki syndrome can include: arched and broad eyebrows, long

palpebral fissures with eversion of the lateral part of lower eyelids, prominent or cupped ears, and a depressed nasal tip and short columella, as well as other findings.



Noonan syndrome

Facial characteristics of Noonan syndrome can include: widely spaced eyes, low-set and posteriorly angulated ears, ptosis, downslanted palpebral fissures, and epicanthal folds, as well as other findings.



Williams syndrome

Facial characteristics of Williams syndrome can include: periorbital fullness, flat nasal bridge, small nose with upturned nasal tip, long philtrum, wide mouth, full lips, small chin, as well as other findings.

Faces and Phenotypic output Q1 to 3

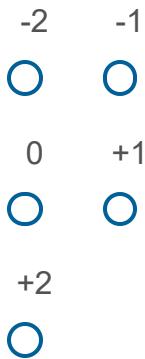
You will now start viewing facial images. First, you will see the image without AI tool outputs. After making a decision for this image, you will be shown our AI tool outputs along with the image and you will be able to make a new prediction based on the additional information. Please note that not all images were correctly classified by the AI tool.

image



Select the genetic condition or unaffected.

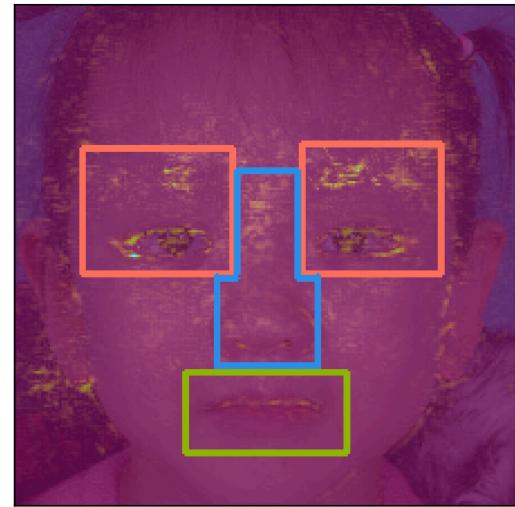
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



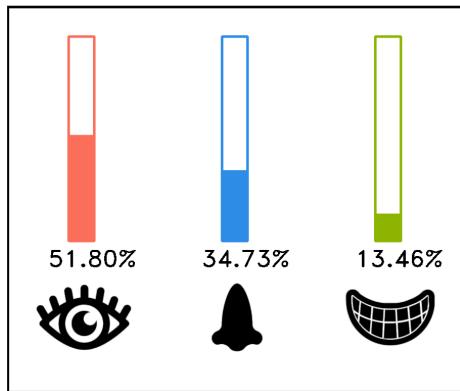
image



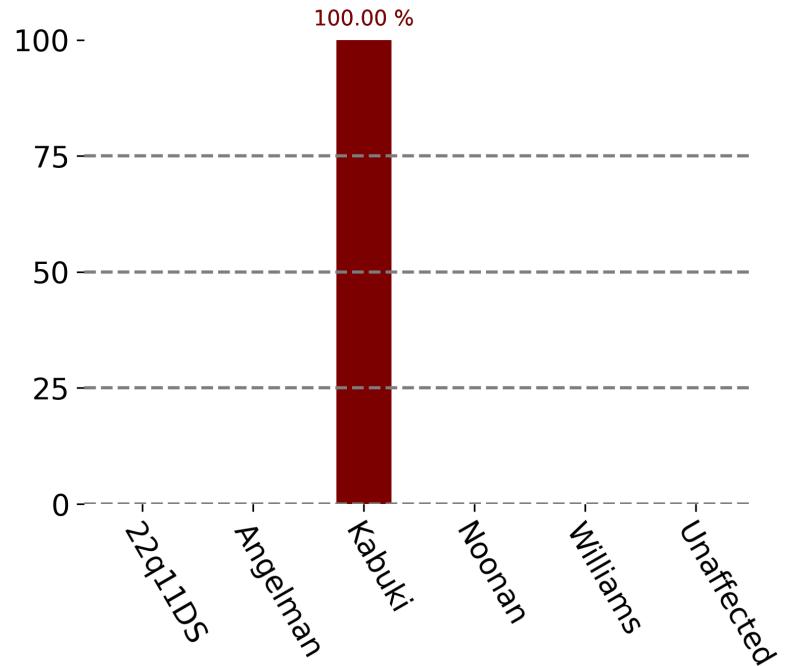
saliency map



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

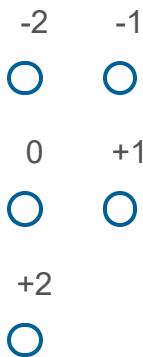


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map



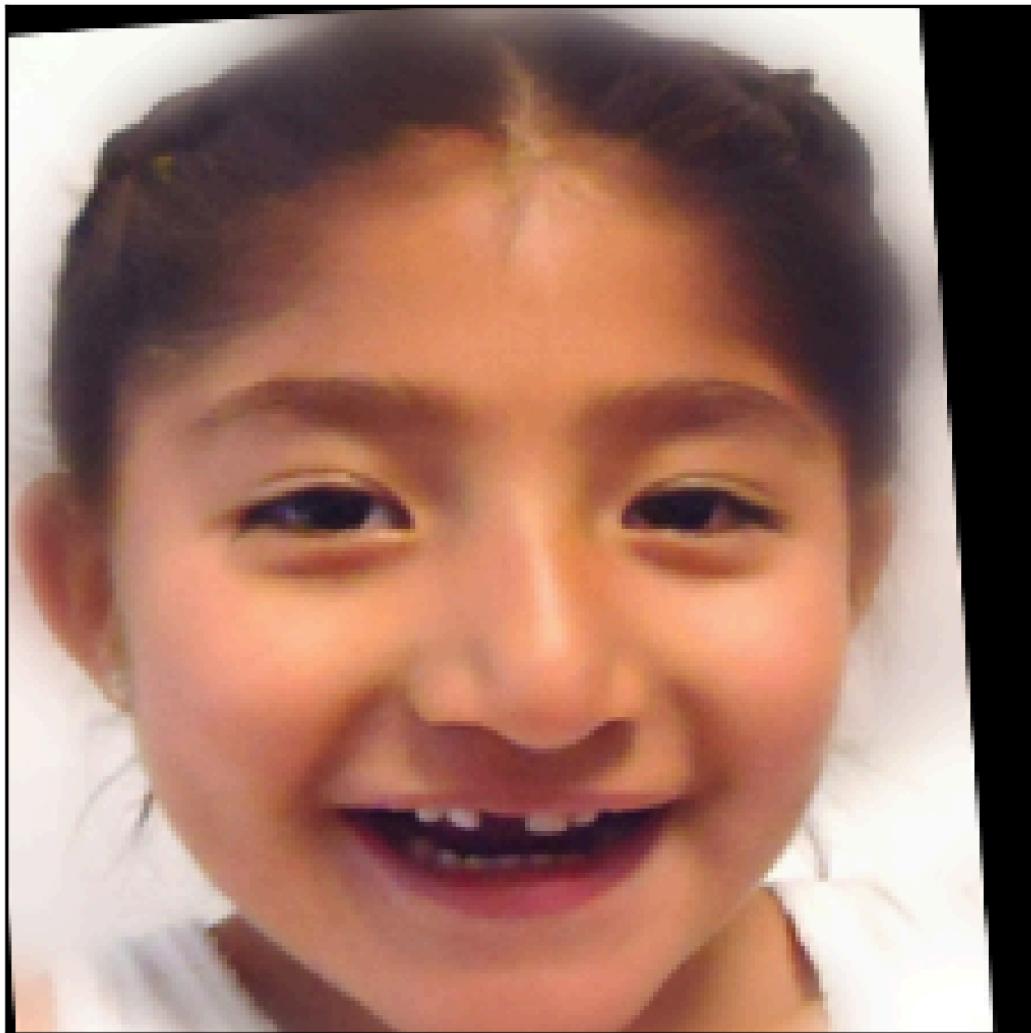
Region relevance



Prediction probability

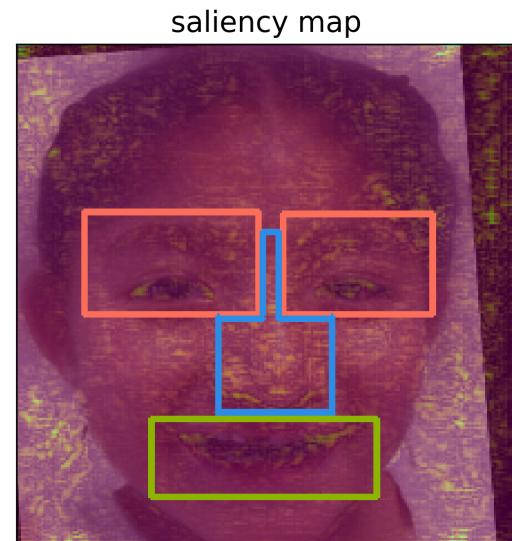
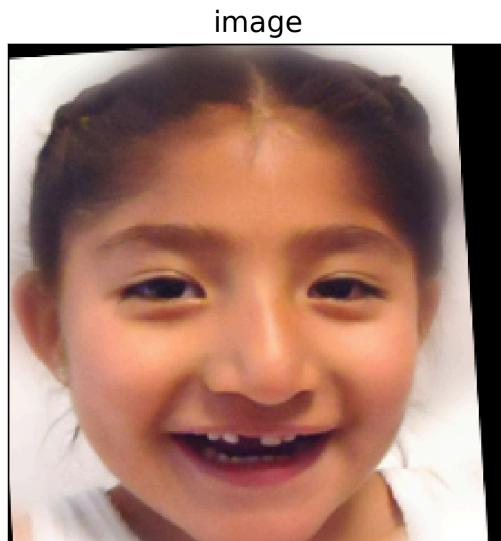
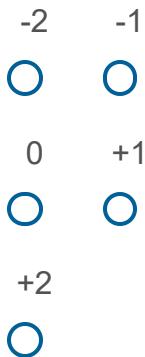
-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

image

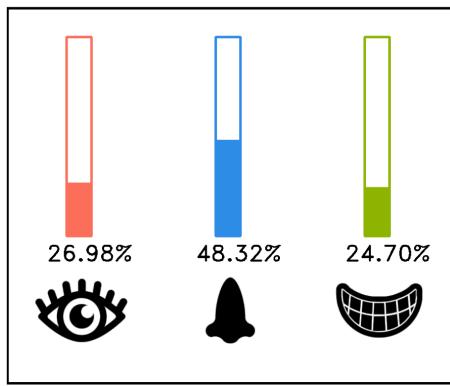


Select the genetic condition or unaffected.

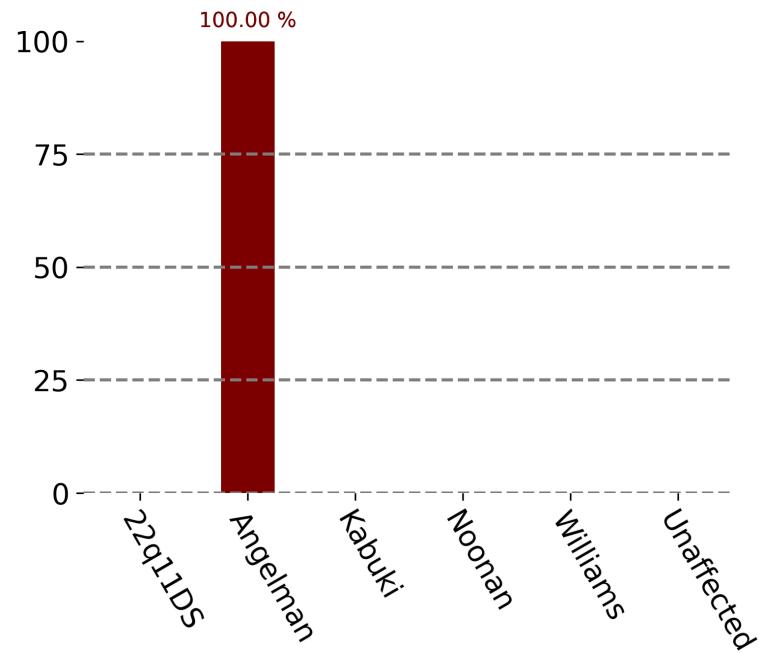
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2 -1

0 +1

+2

How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map

-2 -1

0 +1

+2

Region relevance

-2 -1

0 +1

+2



Prediction probability

-2 -1



0 +1

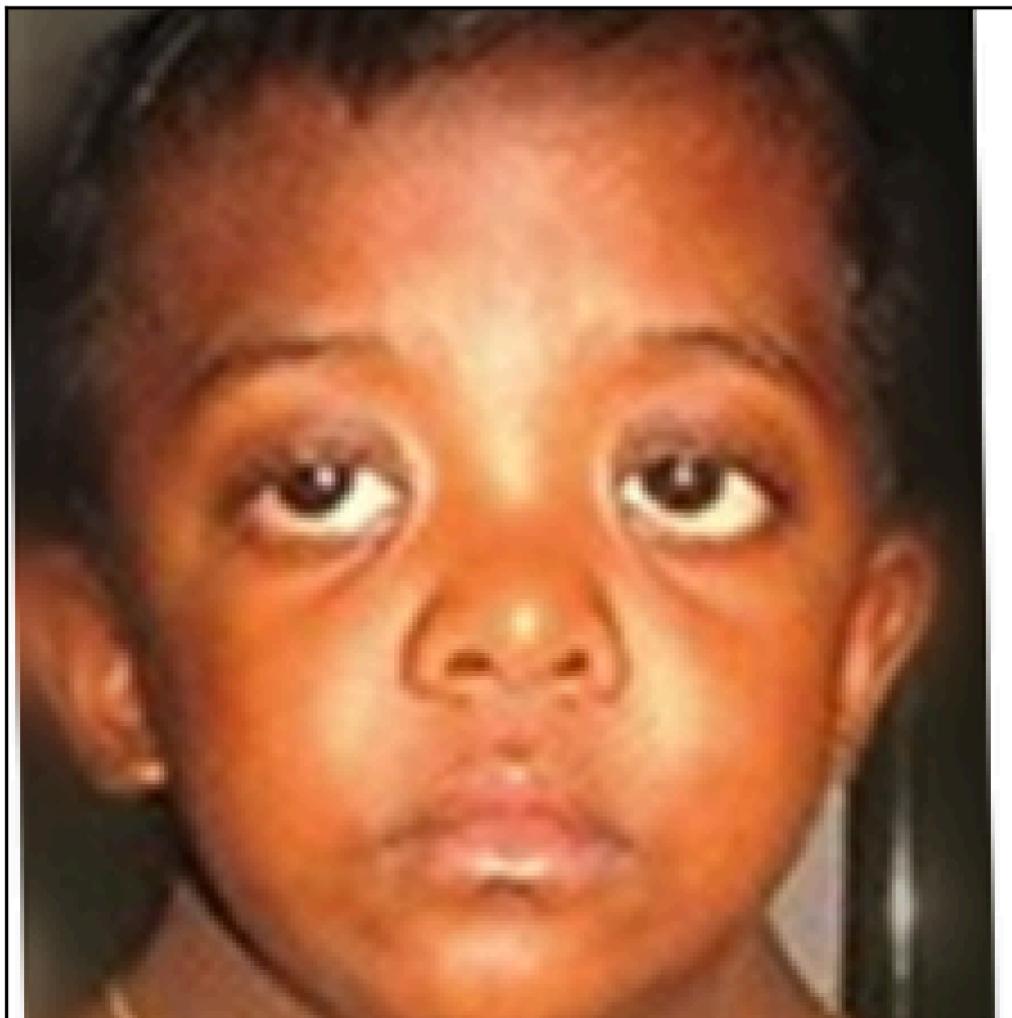


+2



Faces and Phenotypic output Q4 to 6

image

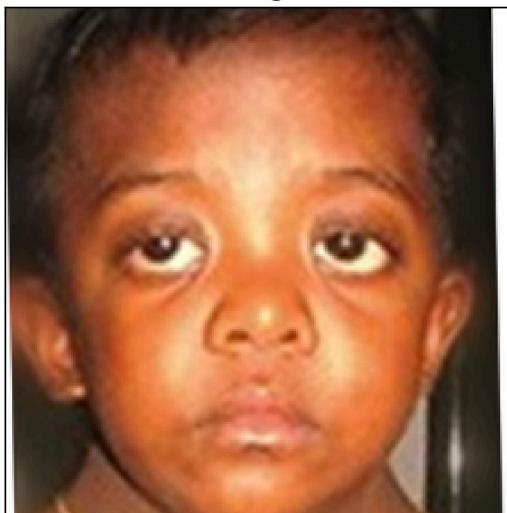


Select the genetic condition or unaffected.

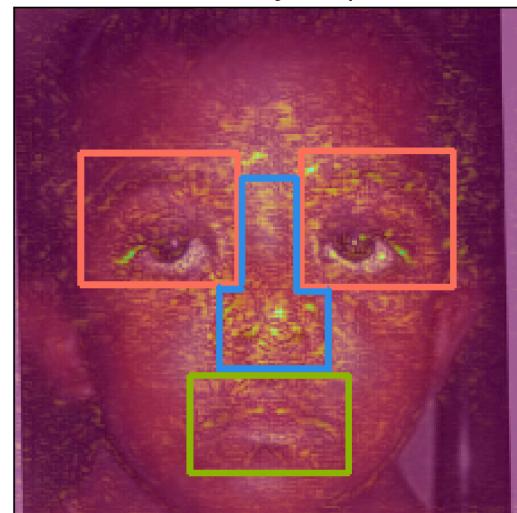
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2	-1
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0	+1
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+2	
<input type="radio"/>	

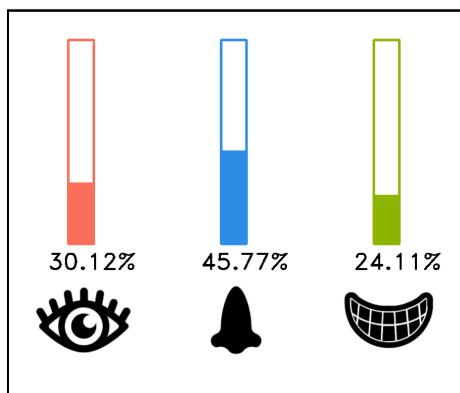
image



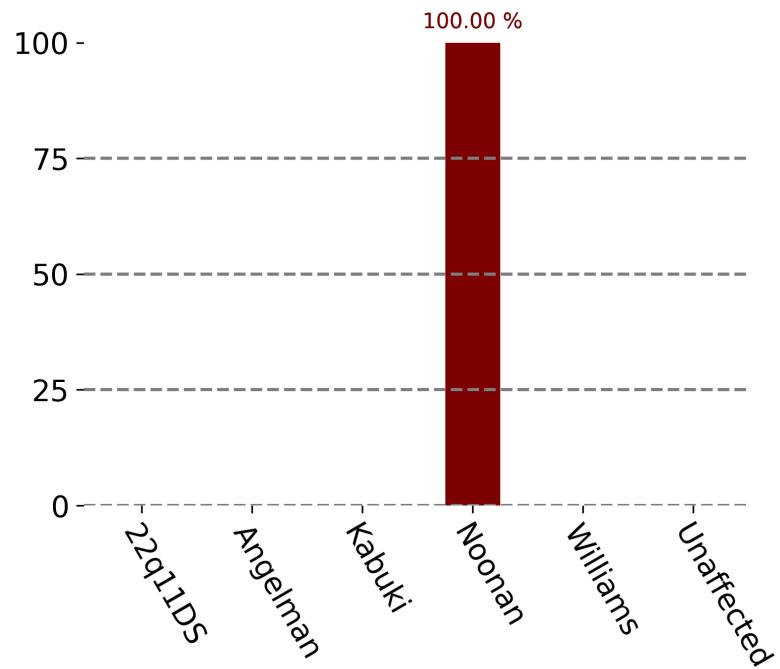
saliency map



region relevance



classifier probabilities



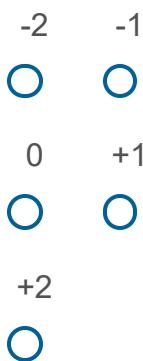
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

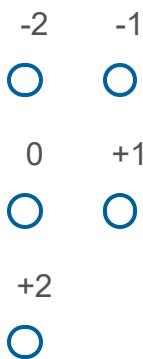


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

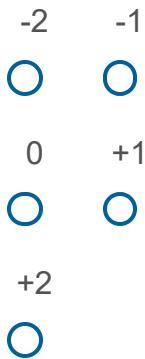
Saliency map



Region relevance



Prediction probability

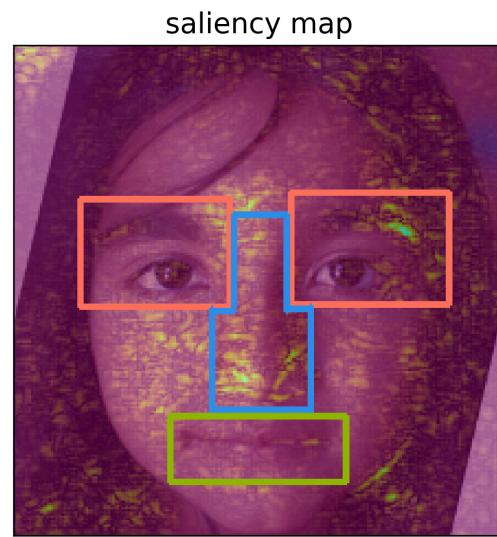


image

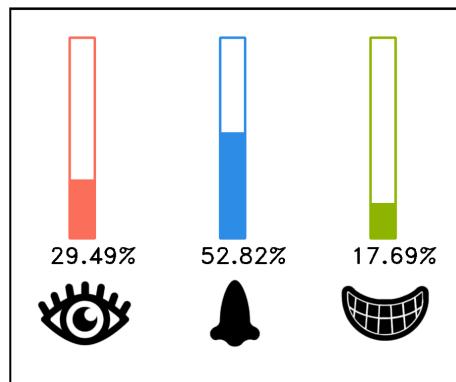


Select the genetic condition or unaffected.

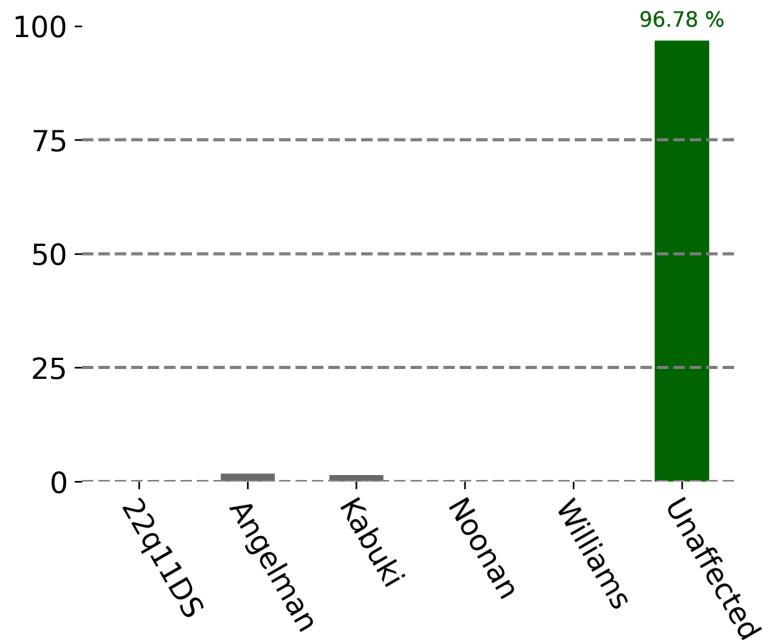
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2 -1

0 +1

+2

How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map

-2 -1

0 +1

+2

Region relevance

-2 -1

0 +1

+2



Prediction probability

-2 -1



0 +1

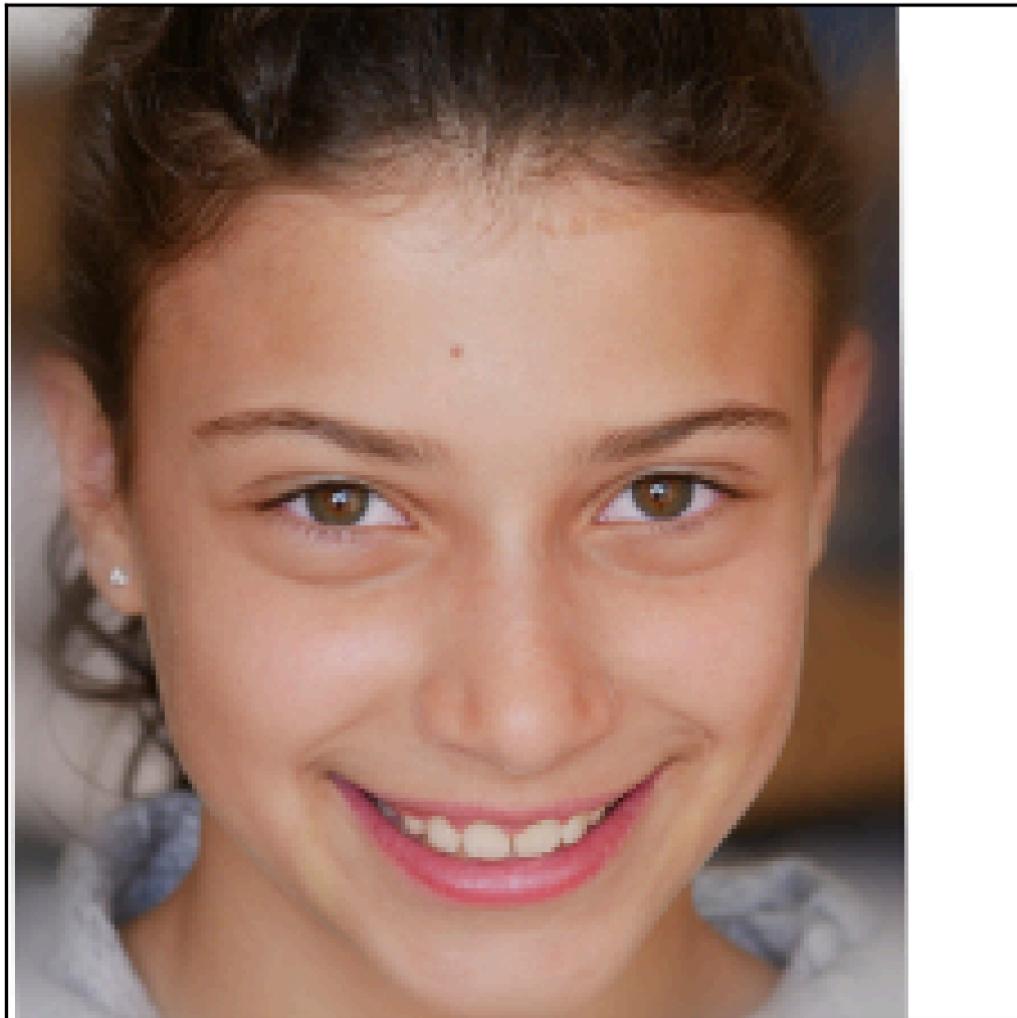


+2



Faces and Phenotypic output Q7 to 9

image

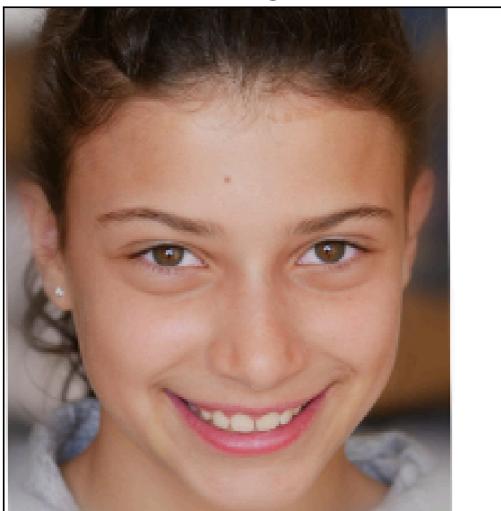


Select the genetic condition or unaffected.

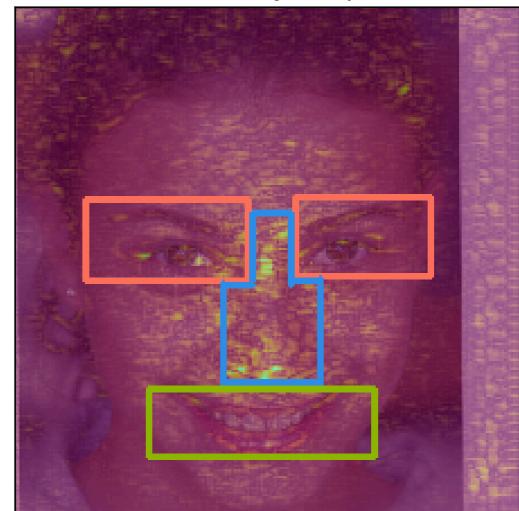
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



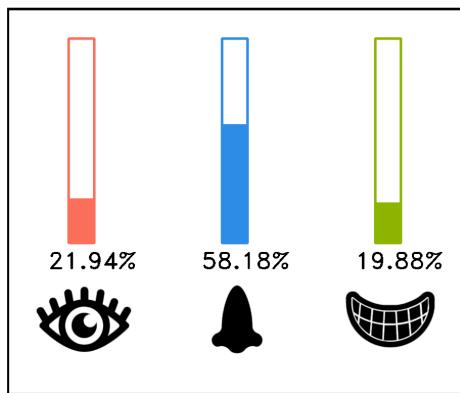
image



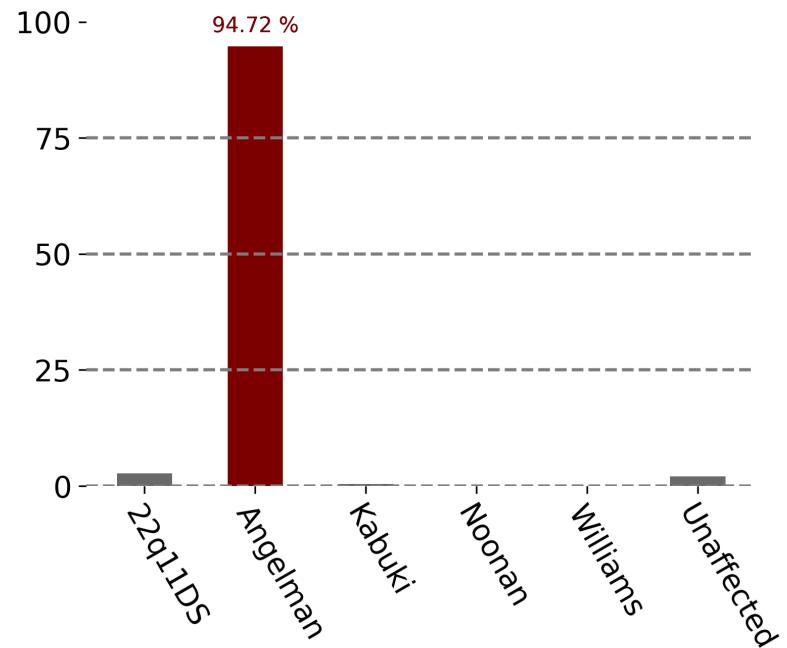
saliency map



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

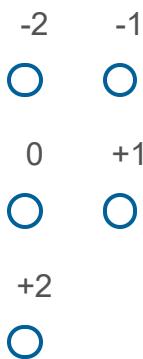


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

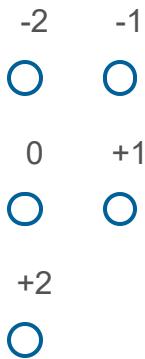
Saliency map



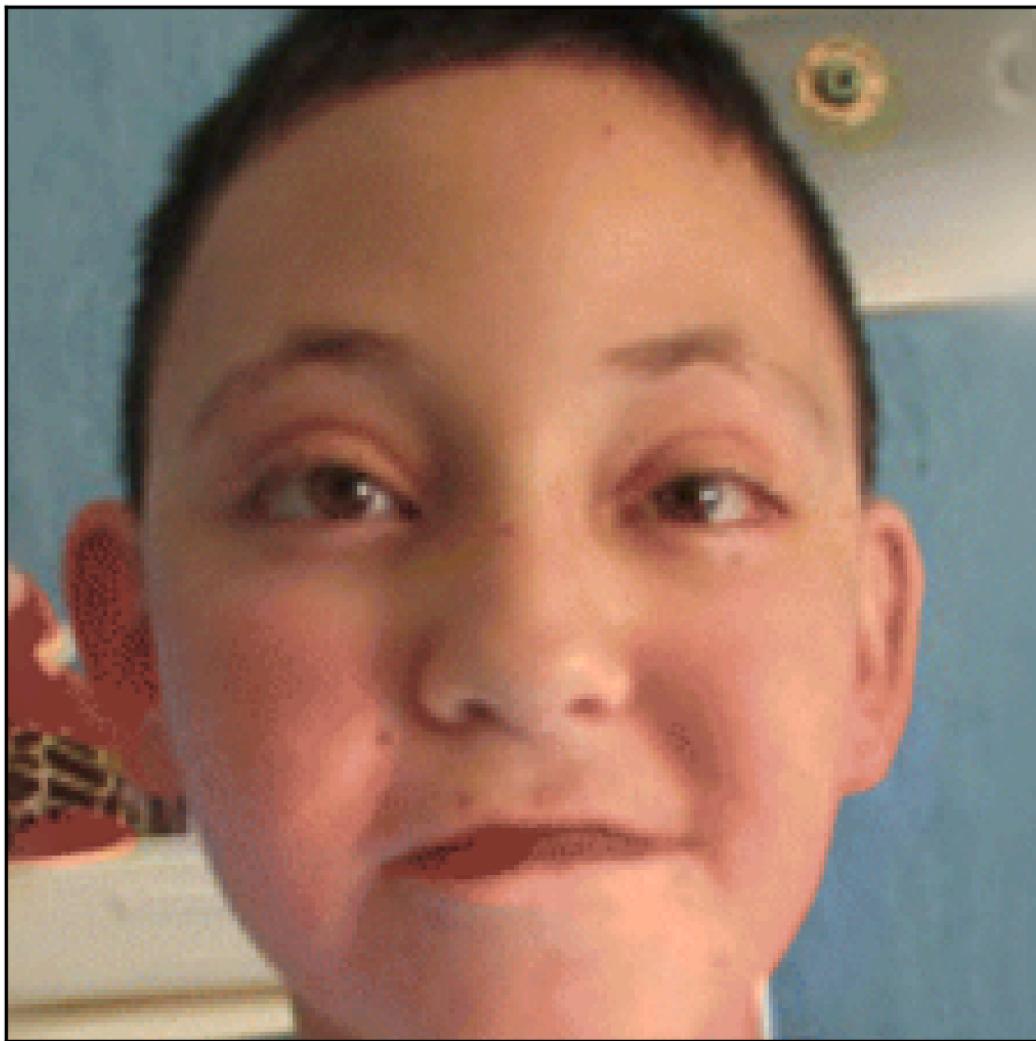
Region relevance



Prediction probability



image

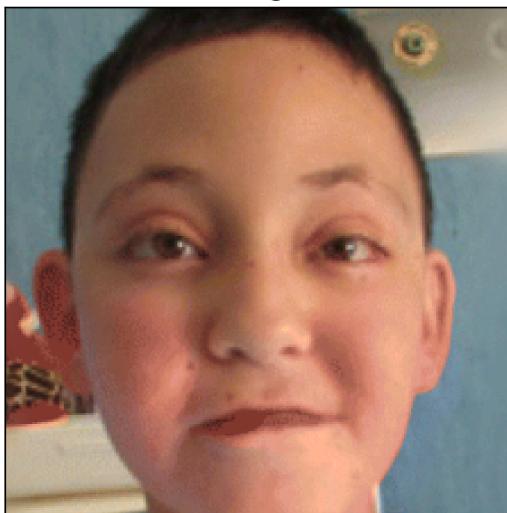


Select the genetic condition or unaffected.

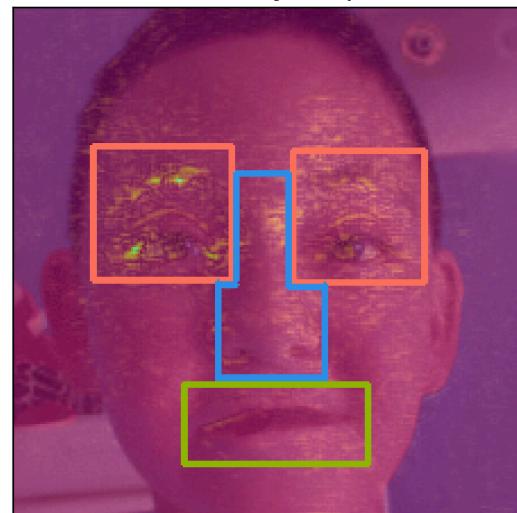
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

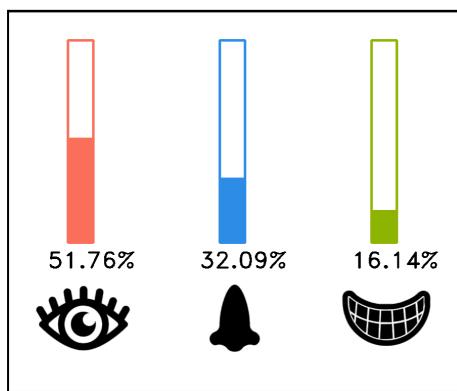
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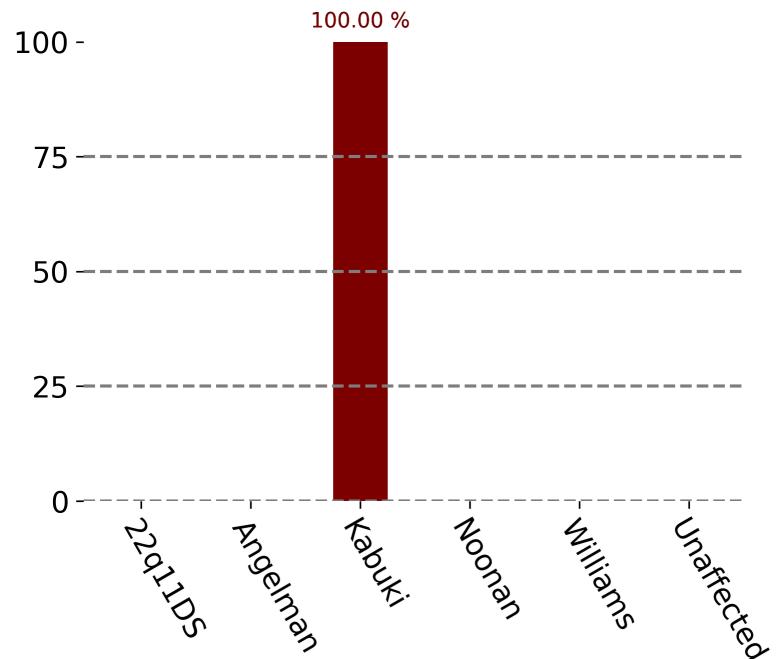
saliency map



region relevance



classifier probabilities



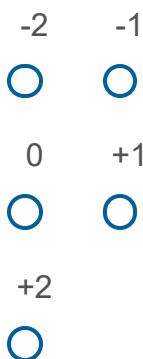
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

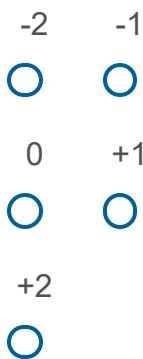


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map



Region relevance



Prediction probability

-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

image



Select the genetic condition or unaffected.

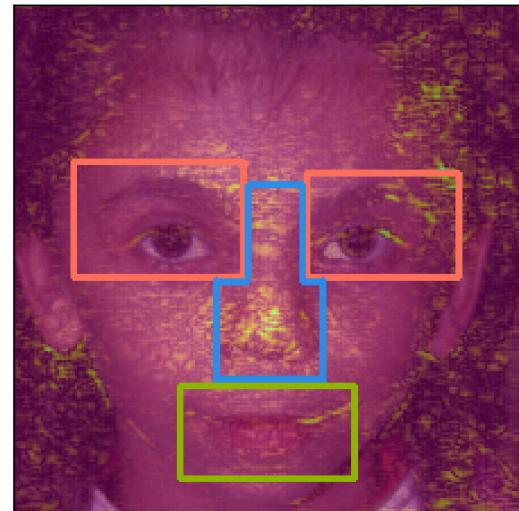
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2	-1
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+2	
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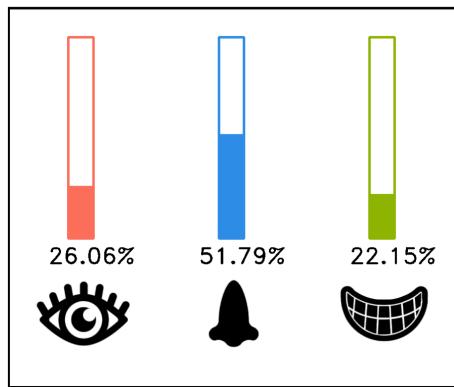
image



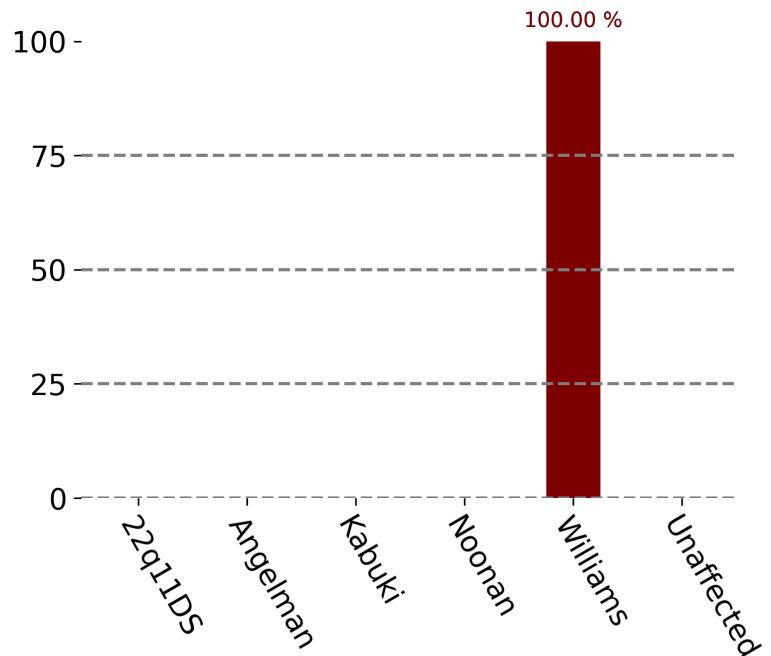
saliency map



region relevance



classifier probabilities



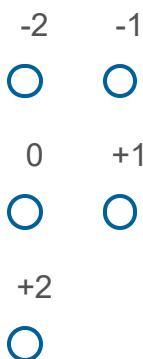
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

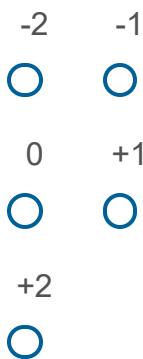


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

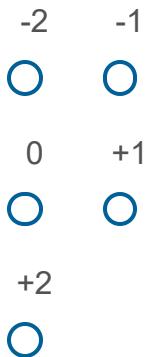
Saliency map



Region relevance

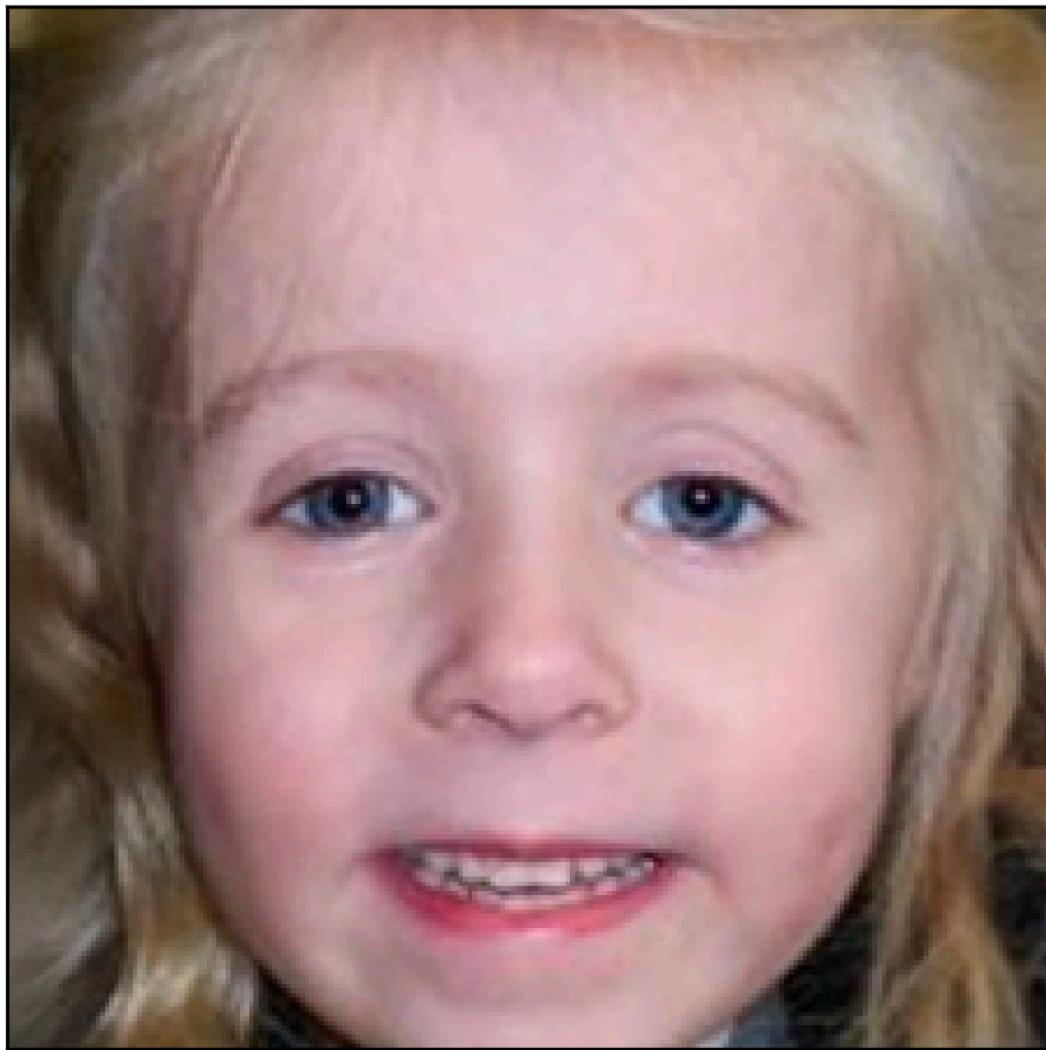


Prediction probability



Faces and Phenotypic output Q10 to 12

image



Select the genetic condition or unaffected.

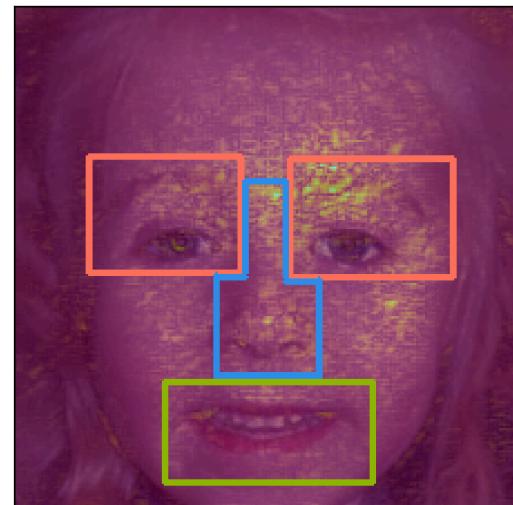
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



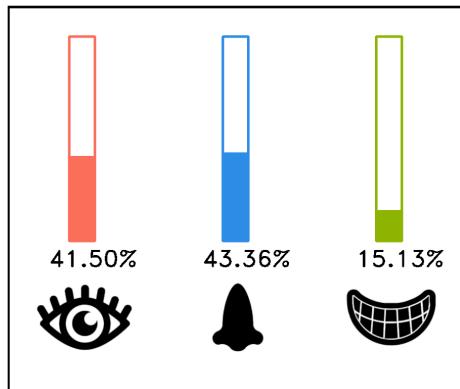
image



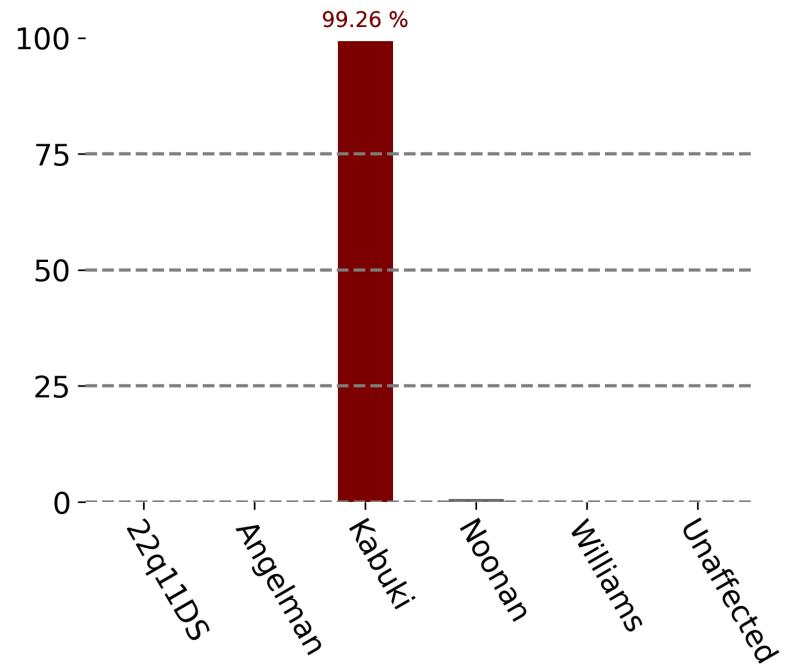
saliency map



region relevance



classifier probabilities



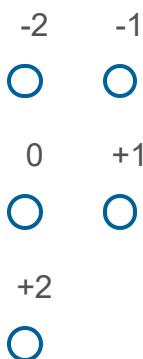
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

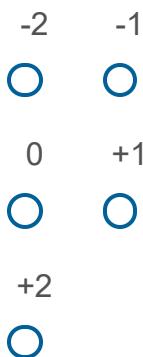


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

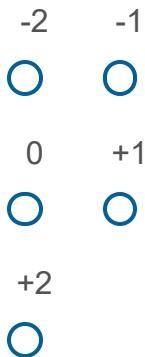
Saliency map



Region relevance

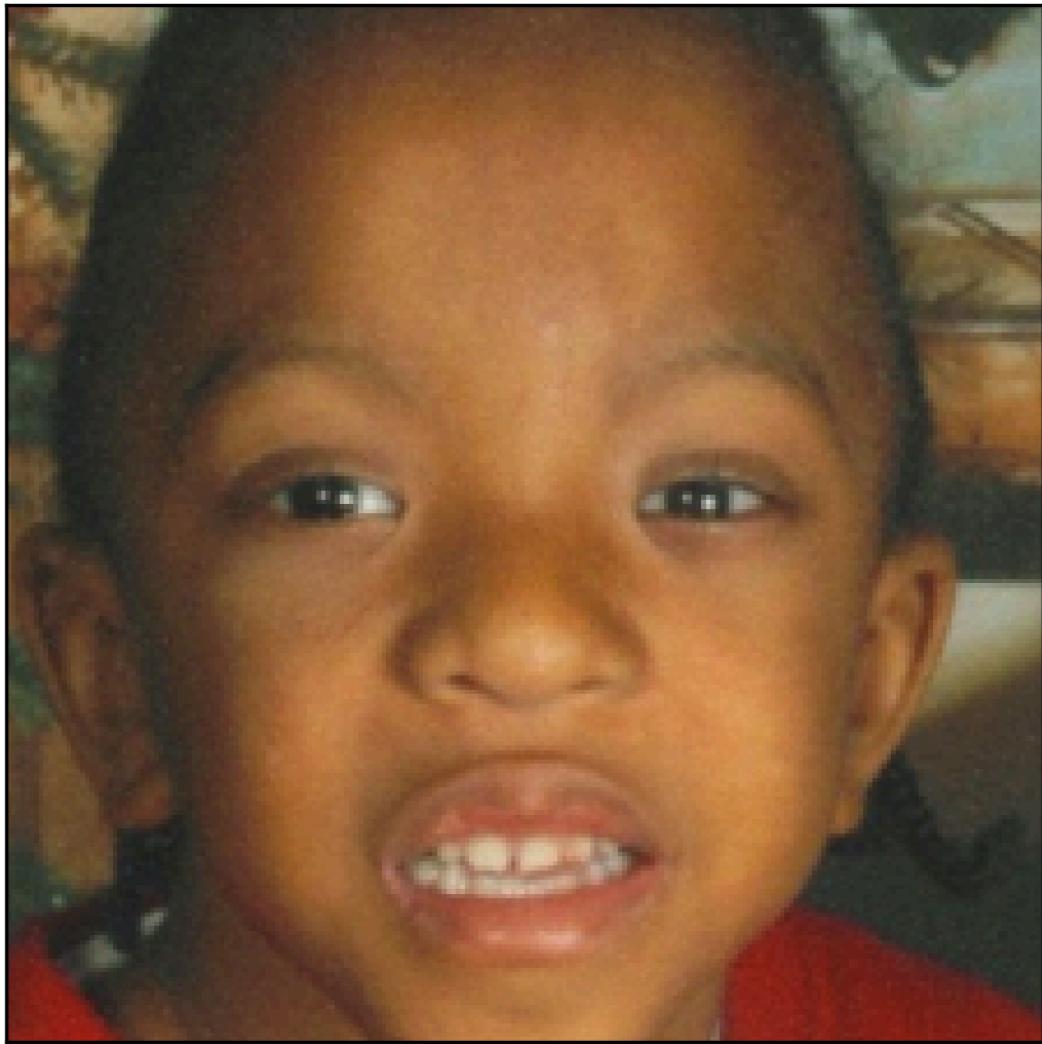


Prediction probability



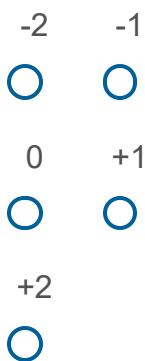
Faces and Phenotypic output Q13 - 15

image

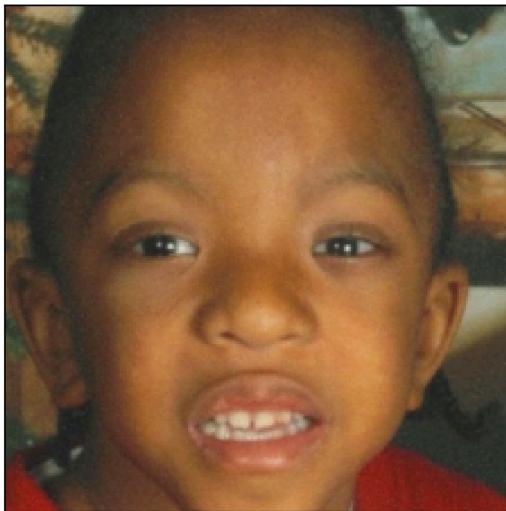


Select the genetic condition or unaffected.

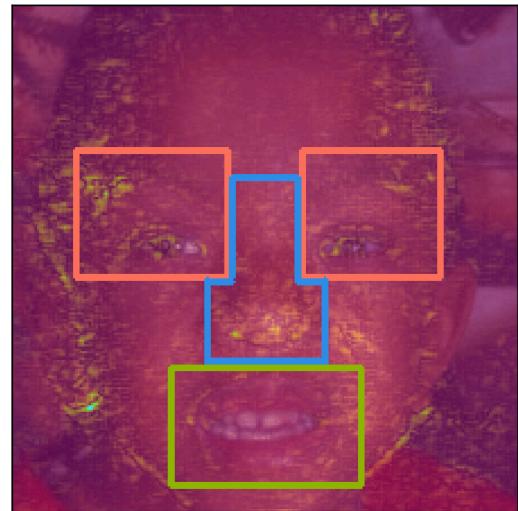
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



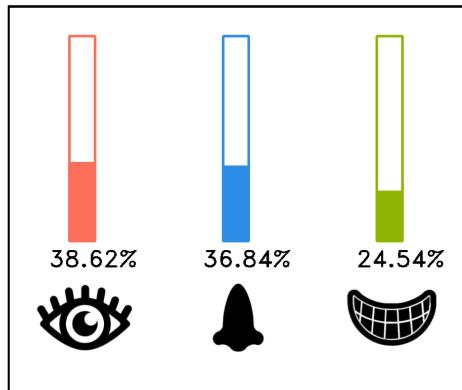
image



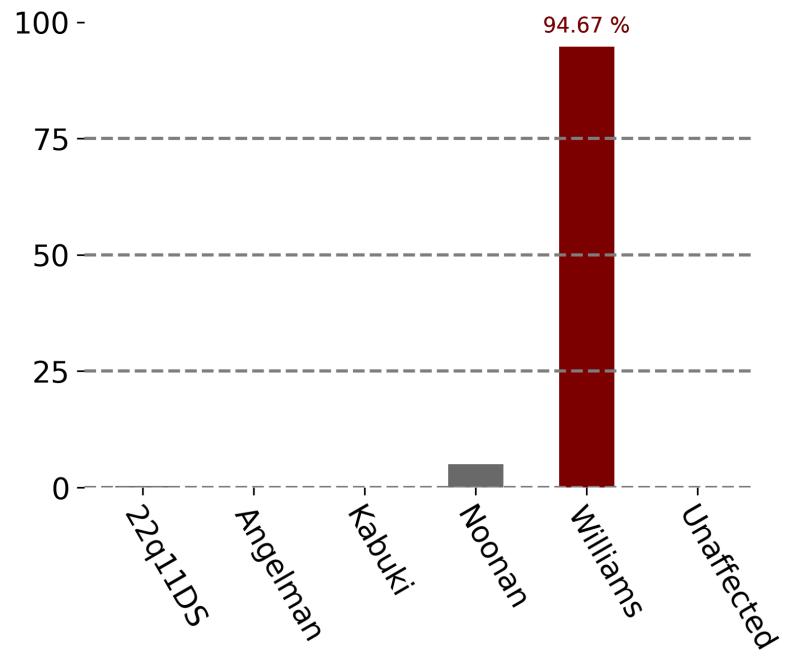
saliency map



region relevance



classifier probabilities



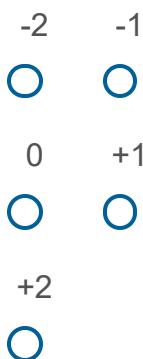
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

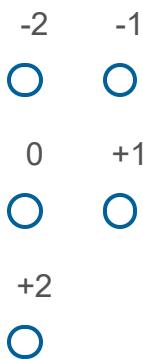


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map



Region relevance



Prediction probability

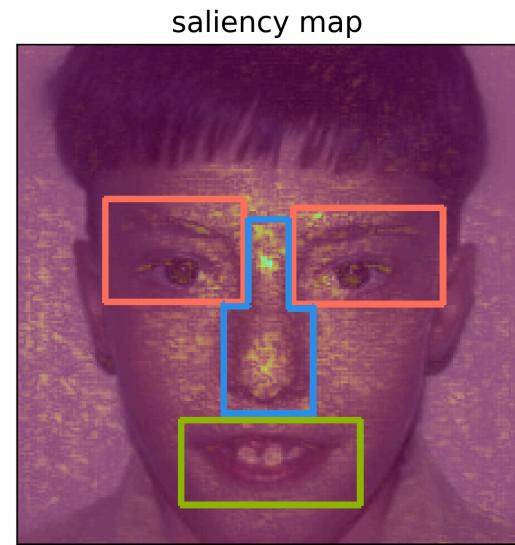
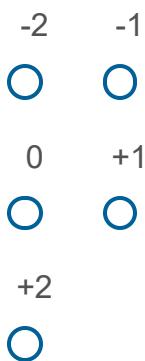
-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

image

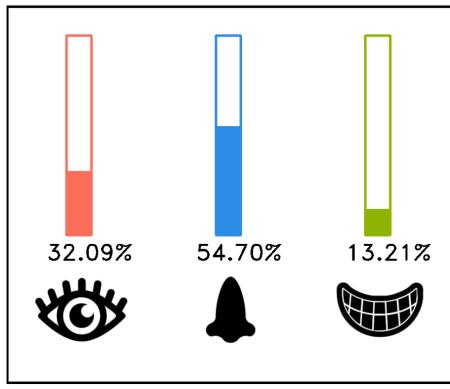


Select the genetic condition or unaffected.

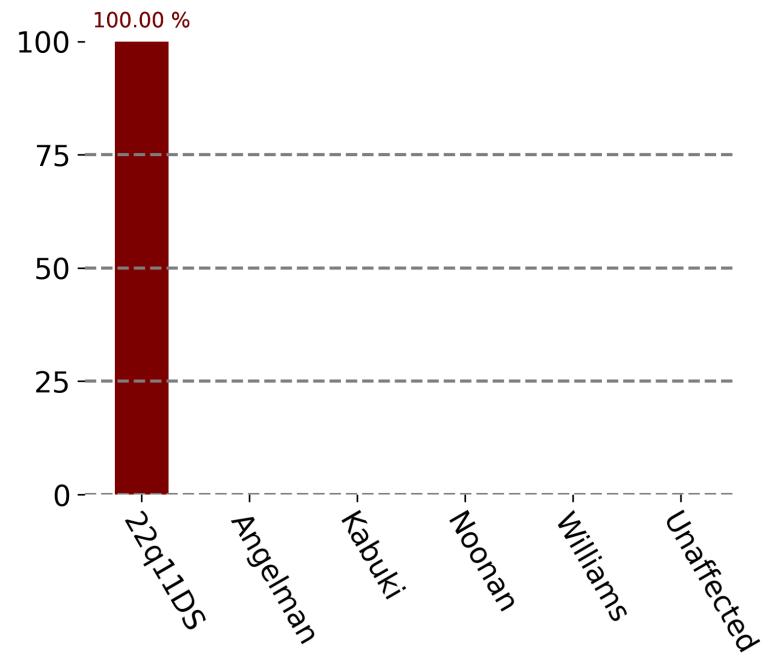
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2 -1

0 +1

+2

How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map

-2 -1

0 +1

+2

Region relevance

-2 -1

0 +1

+2



Prediction probability

-2 -1



0 +1

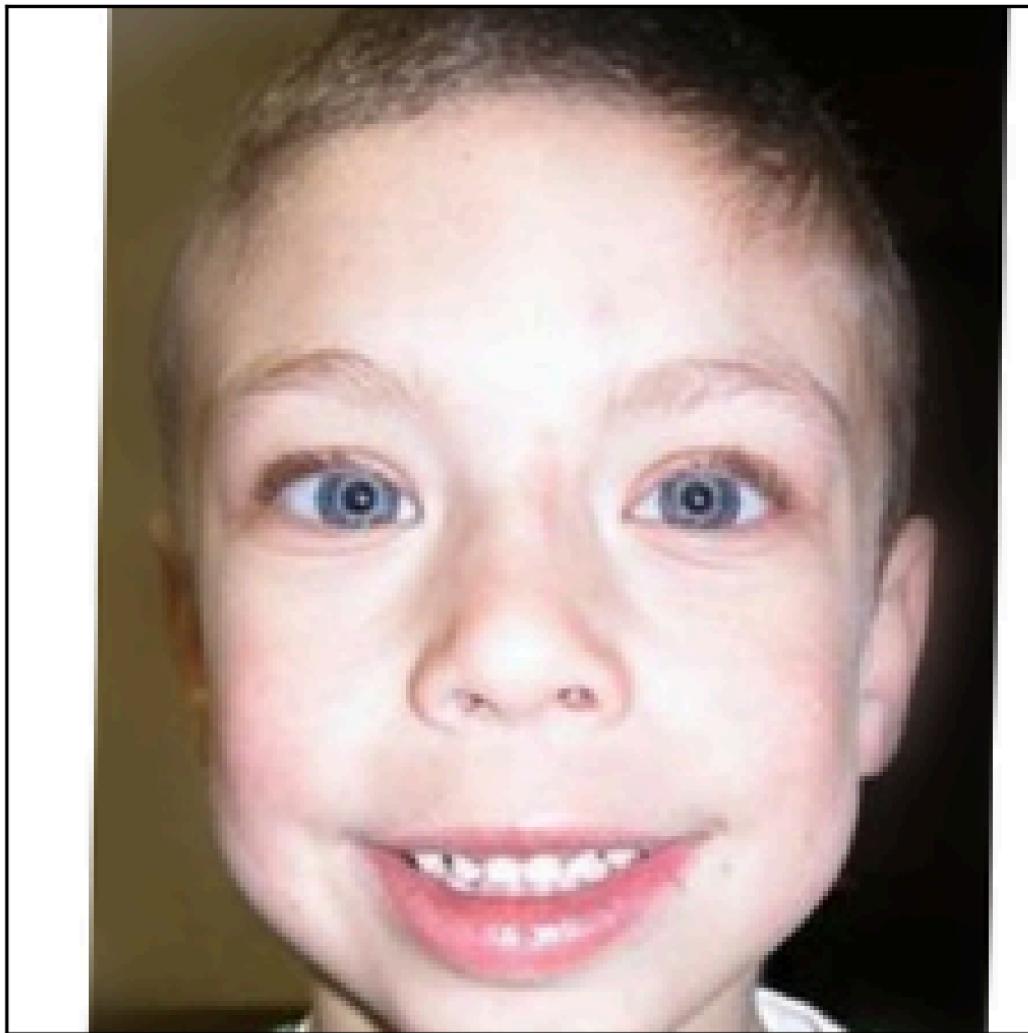


+2



Faces and Phenotypic output Q16 to 18

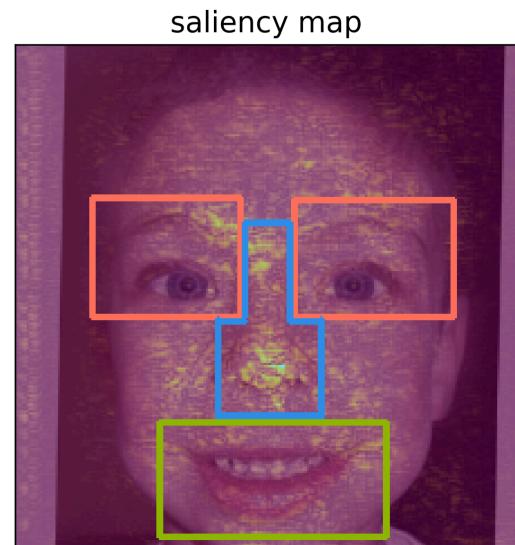
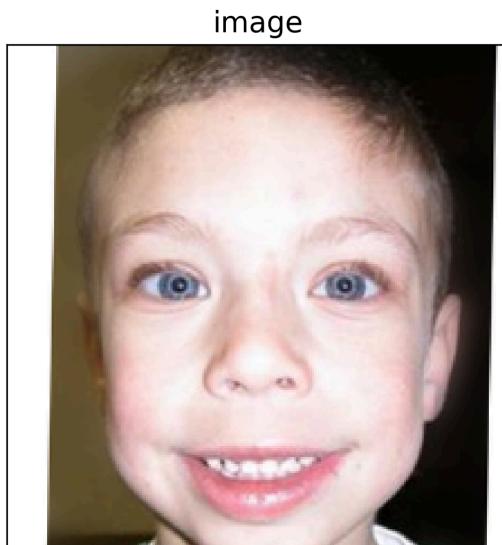
image



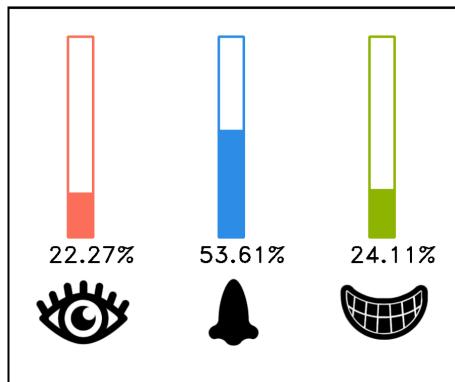
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

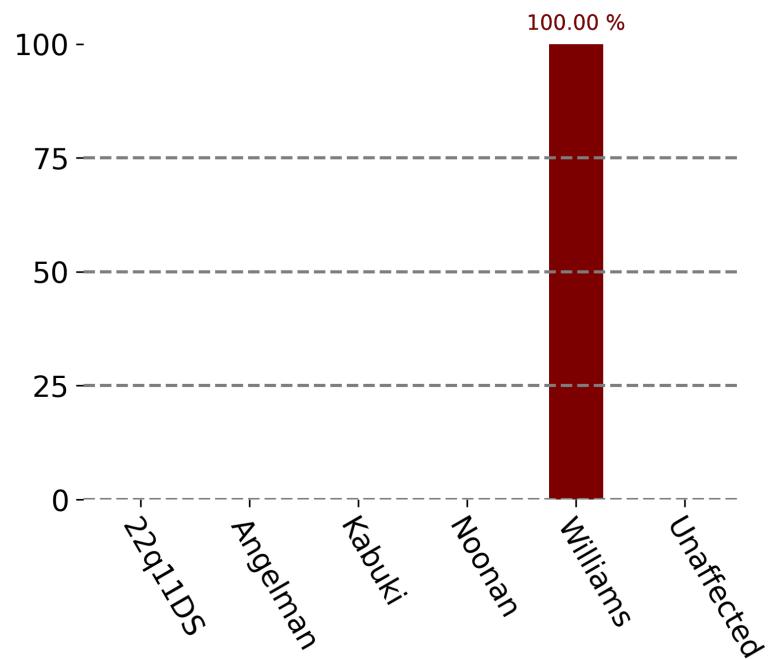




region relevance

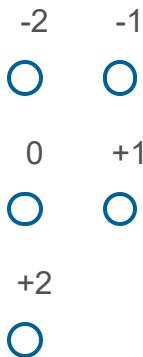


classifier probabilities



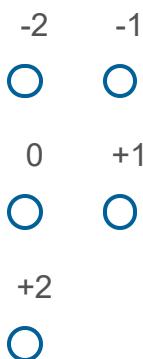
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

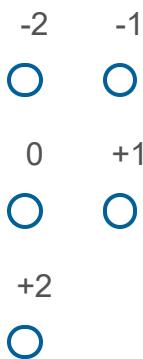


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map



Region relevance



Prediction probability

-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

image



Select the genetic condition or unaffected.

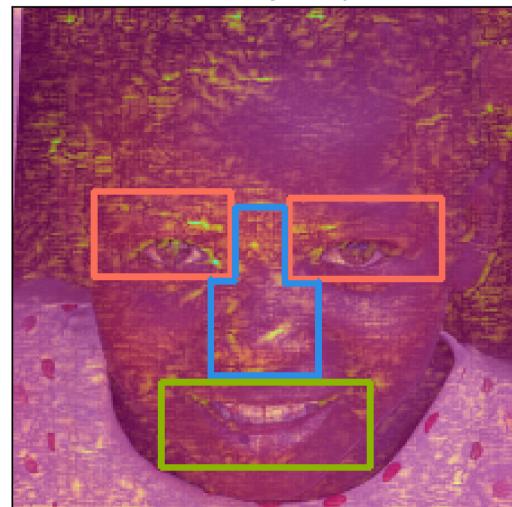
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

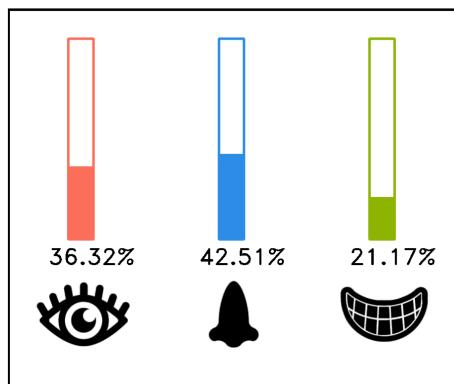
image



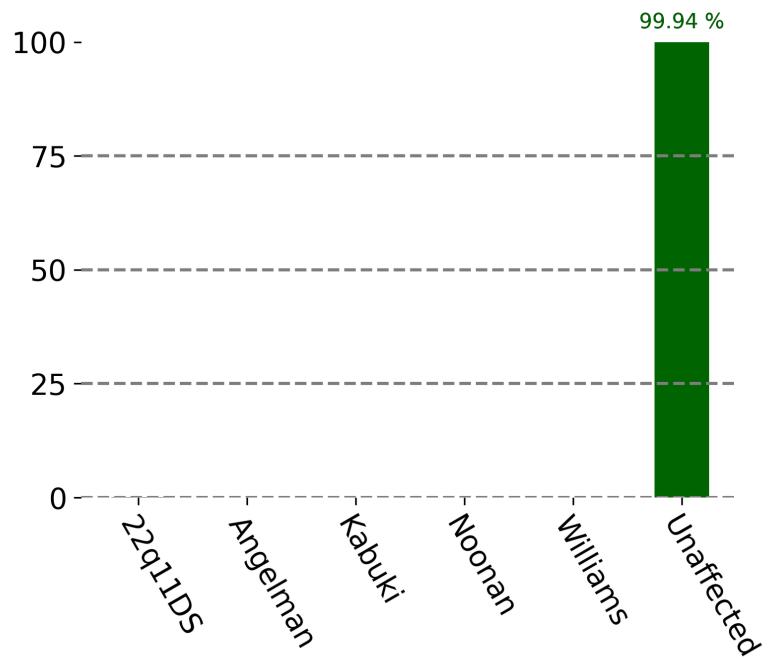
saliency map



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

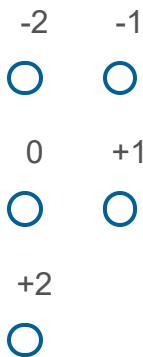


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map



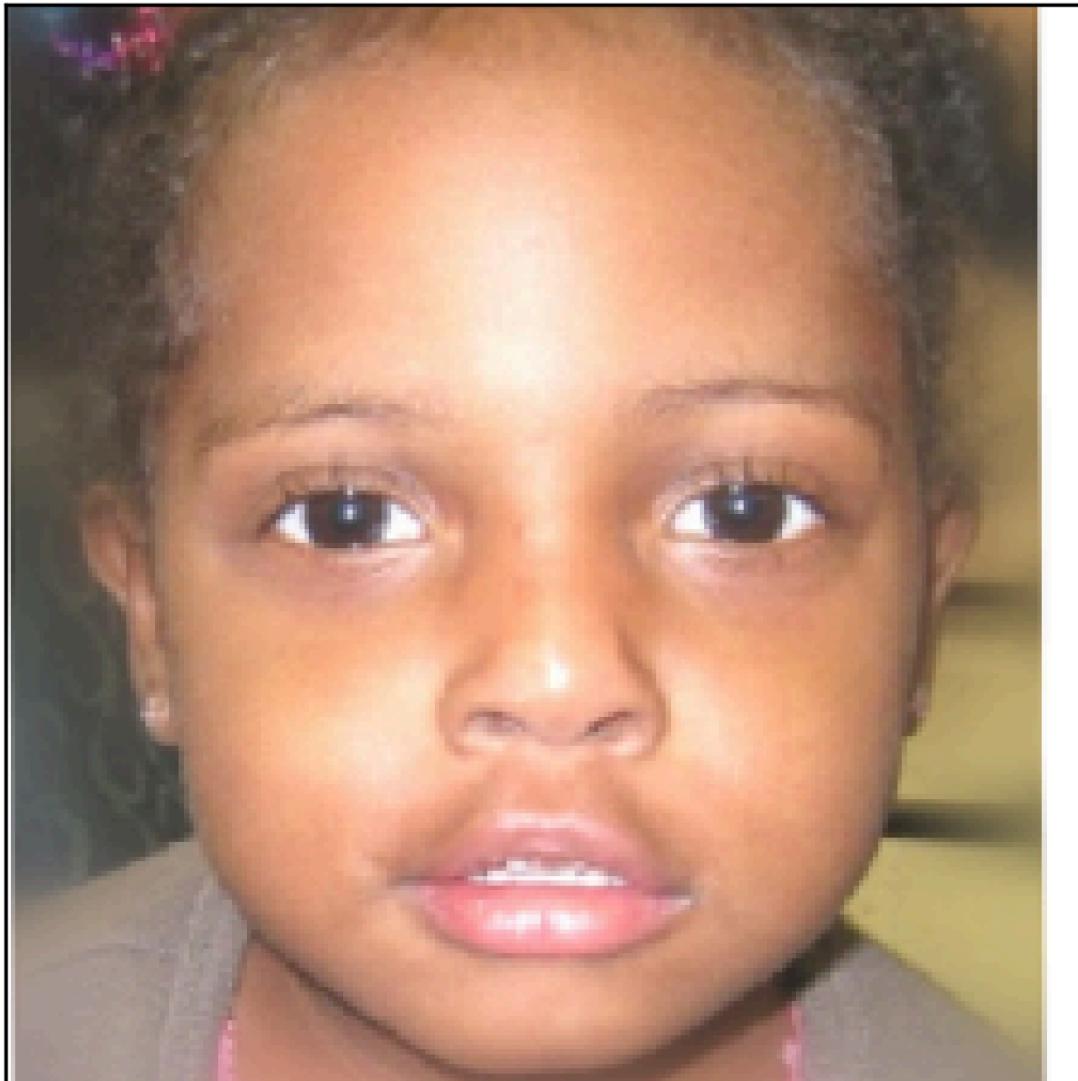
Region relevance



Prediction probability

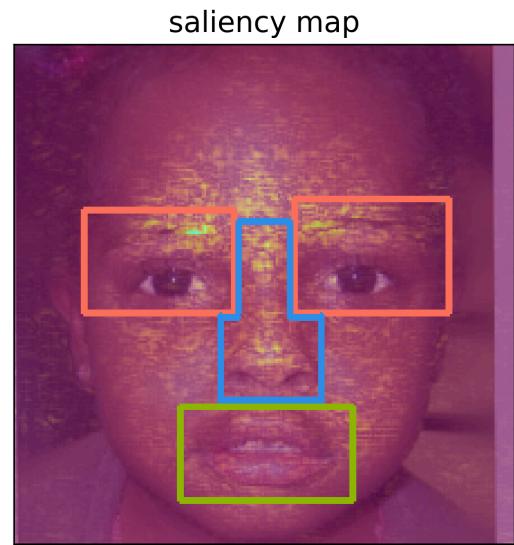
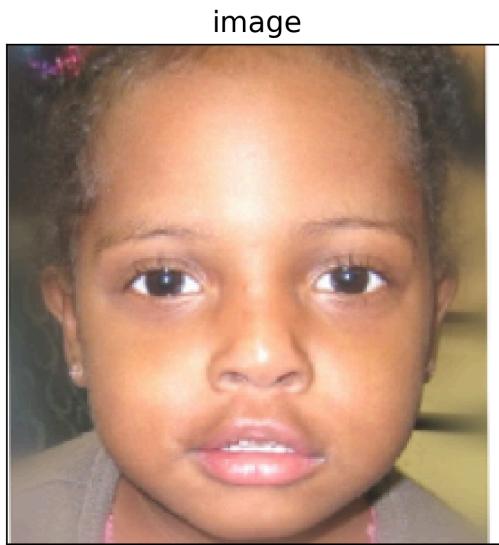
-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

image

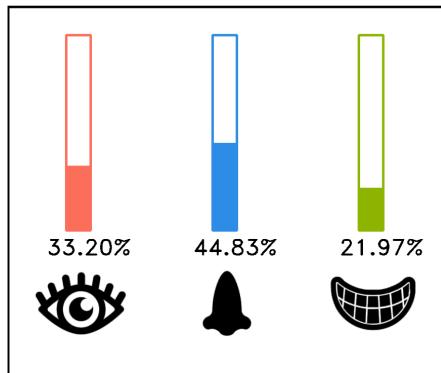


Select the genetic condition or unaffected.

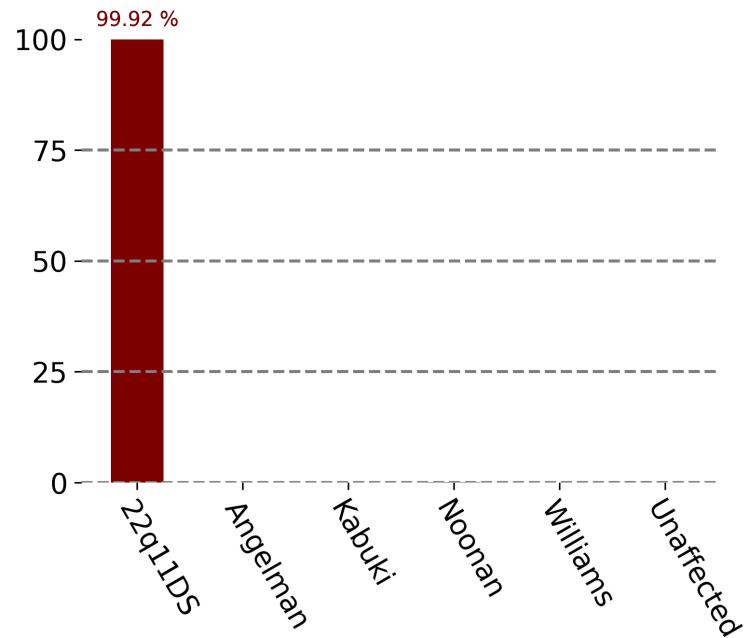
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2 -1

0 +1

+2

How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map

-2 -1

0 +1

+2

Region relevance

-2 -1

0 +1

+2



Prediction probability

-2 -1



0 +1

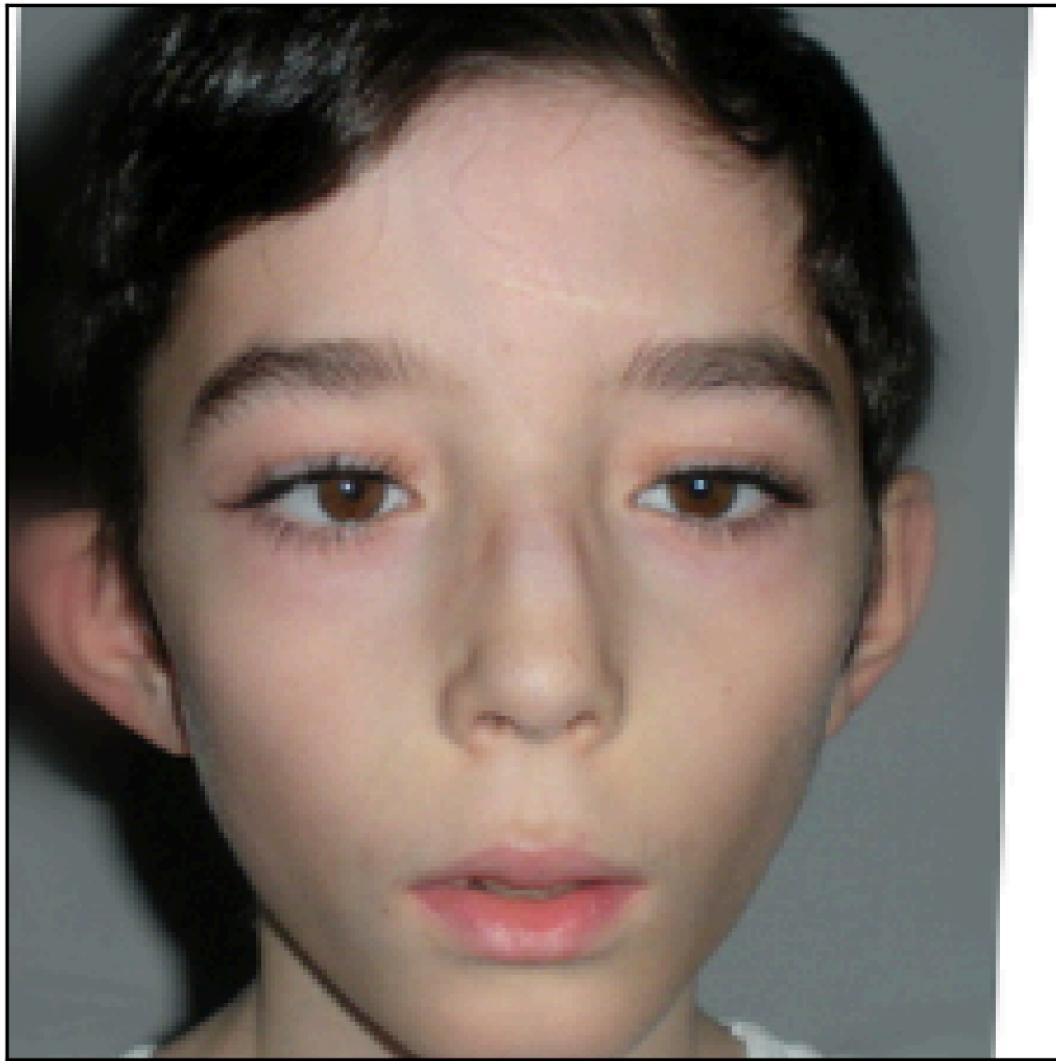


+2



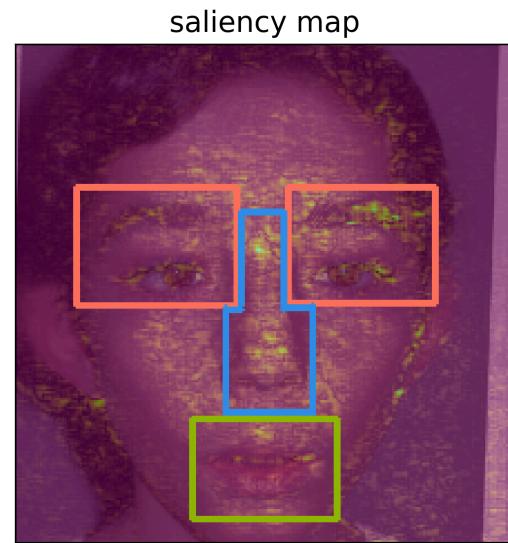
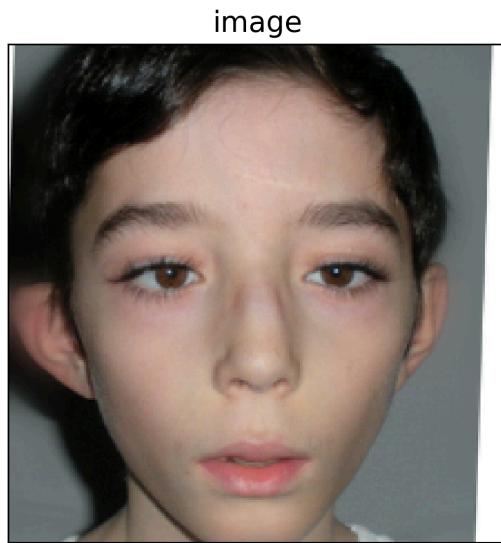
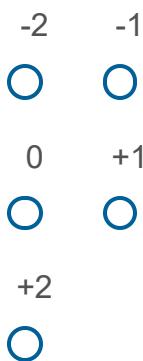
Faces and Phenotypic output Q19 to 21

image

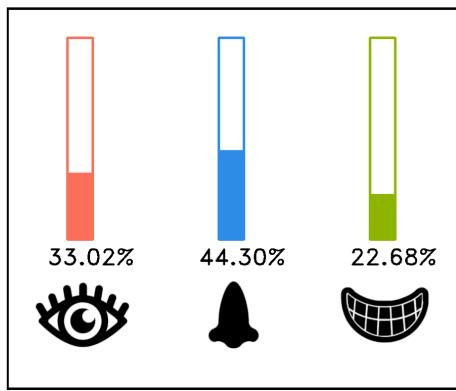


Select the genetic condition or unaffected.

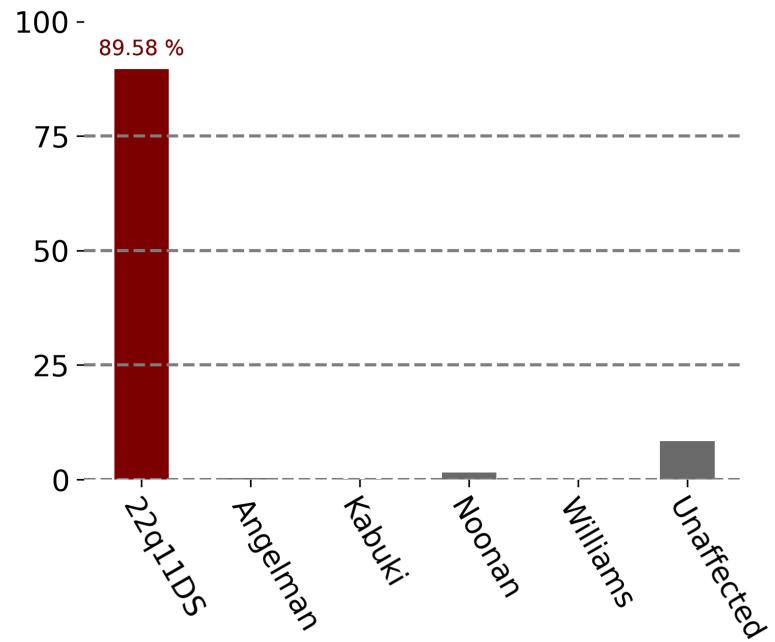
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2 -1

0 +1

+2

How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map

-2 -1

0 +1

+2

Region relevance

-2 -1

0 +1

+2



Prediction probability

-2 -1



0 +1



+2



image



Select the genetic condition or unaffected.

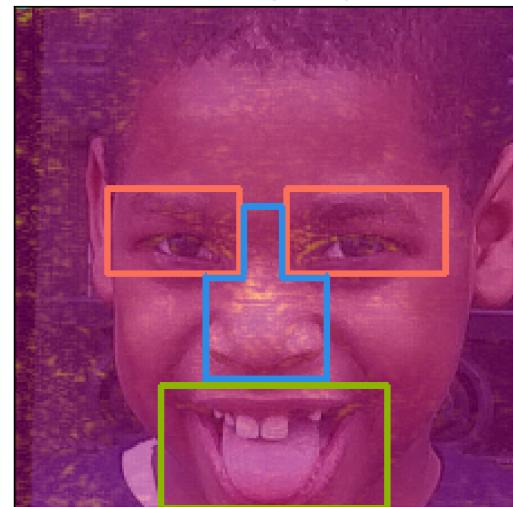
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

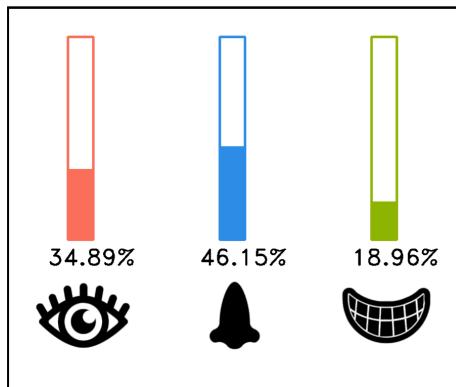
image



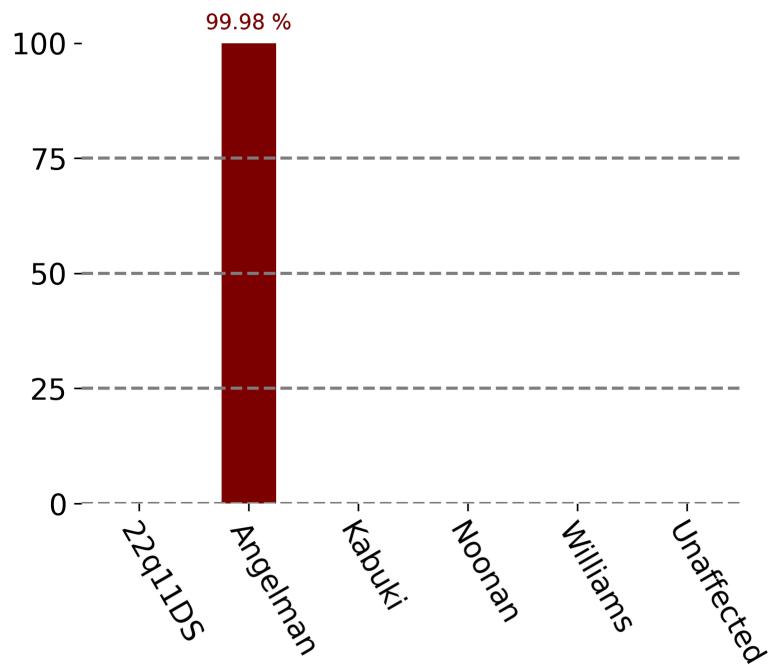
saliency map



region relevance



classifier probabilities



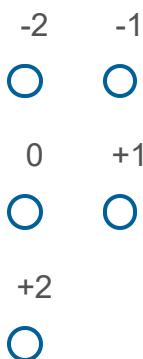
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

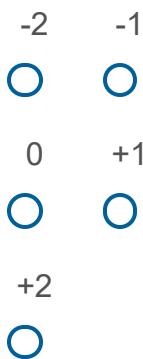


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map



Region relevance



Prediction probability



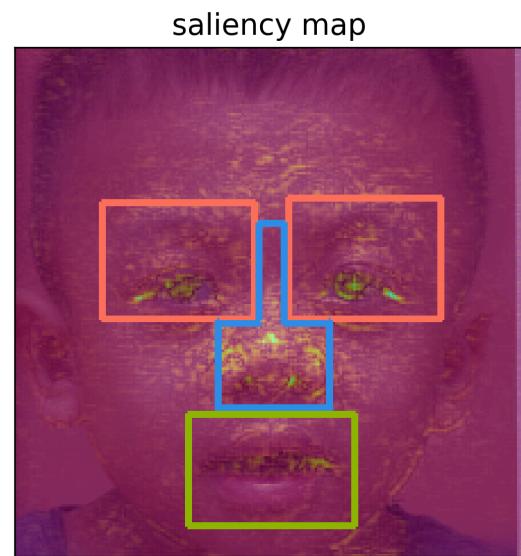
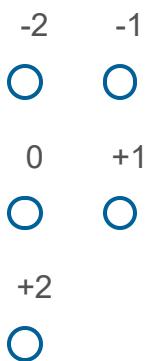
Faces and Phenotypic output Q22 to 24

image

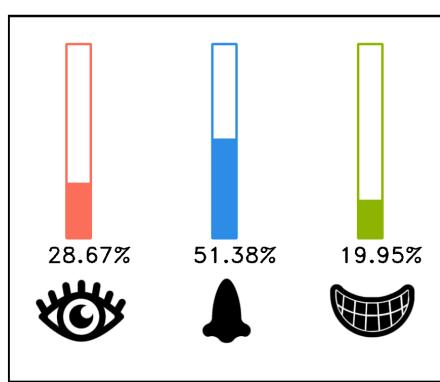


Select the genetic condition or unaffected.

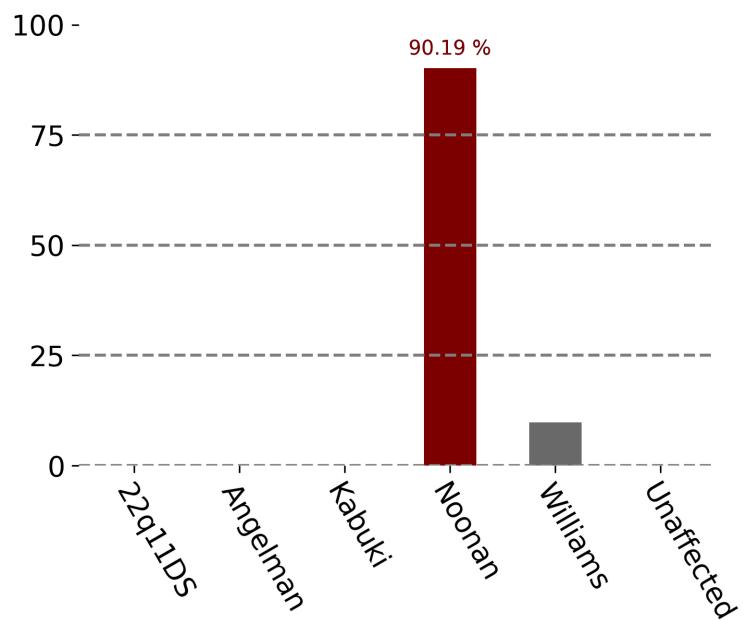
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2 -1

0 +1

+2

How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map

-2 -1

0 +1

+2

Region relevance

-2 -1

0 +1

+2



Prediction probability

-2 -1



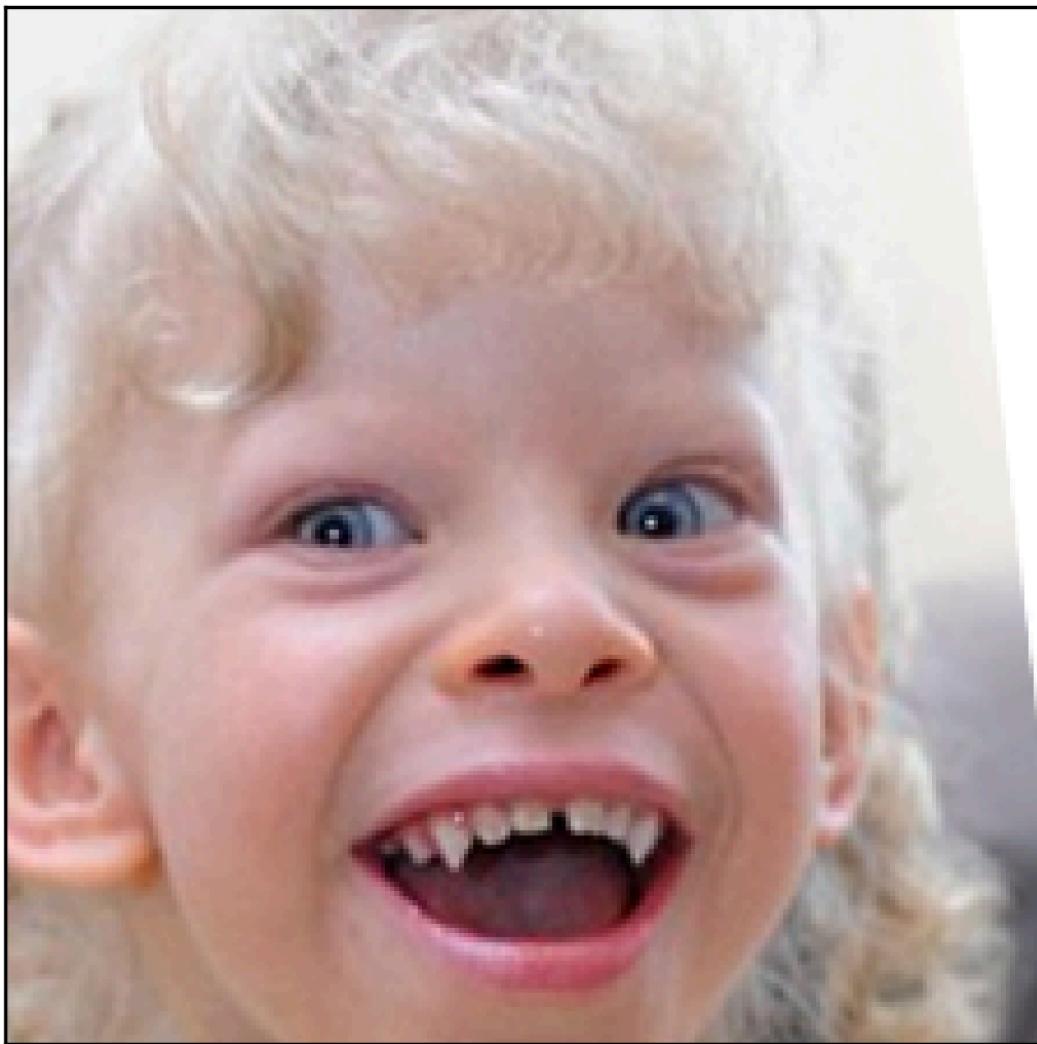
0 +1



+2

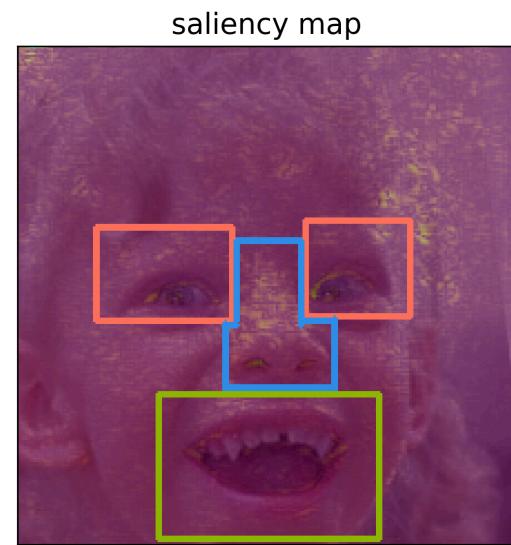
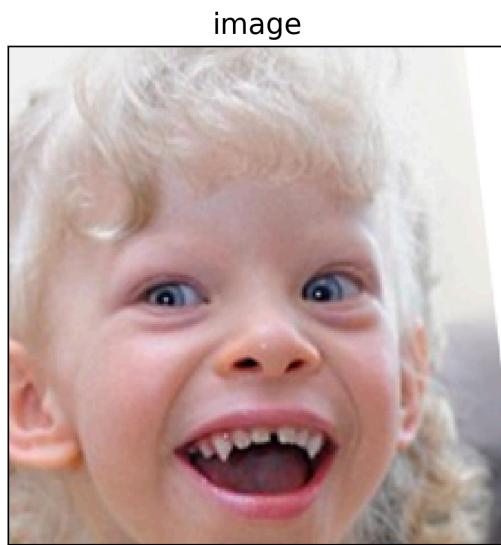


image

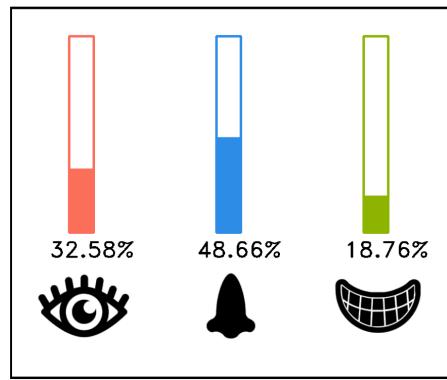


Select the genetic condition or unaffected.

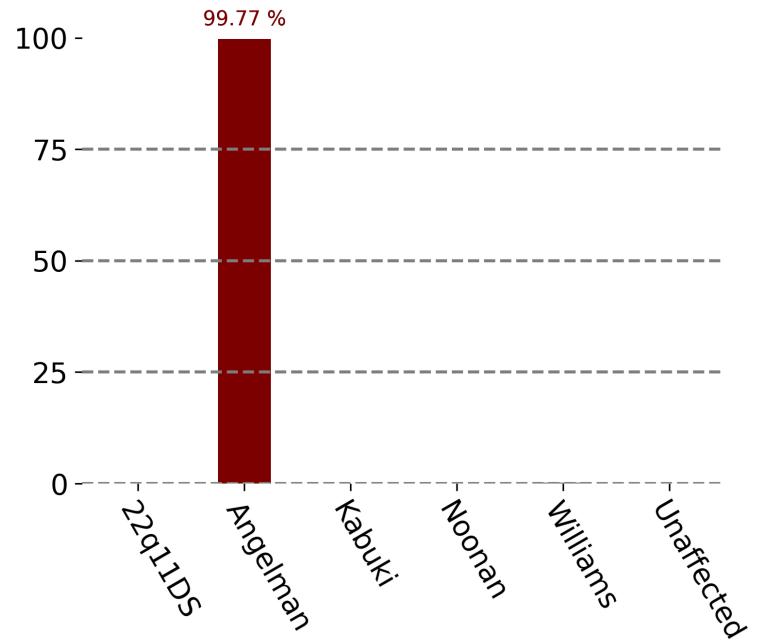
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).



region relevance



classifier probabilities



Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2 -1

0 +1

+2

How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map

-2 -1

0 +1

+2

Region relevance

-2 -1

0 +1

+2



Prediction probability

-2 -1



0 +1



+2



image



Select the genetic condition or unaffected.

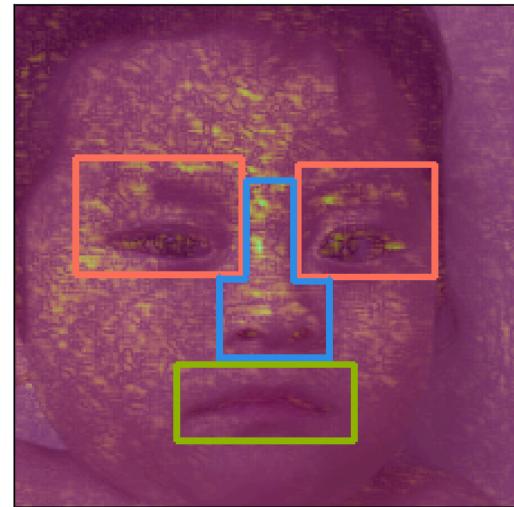
Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

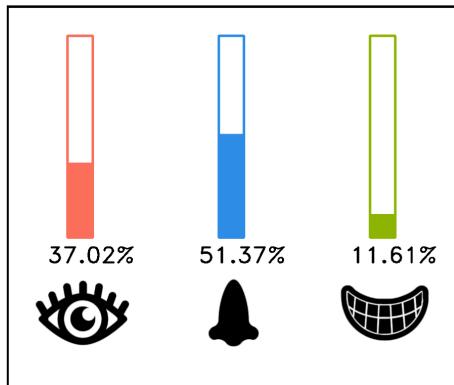
image



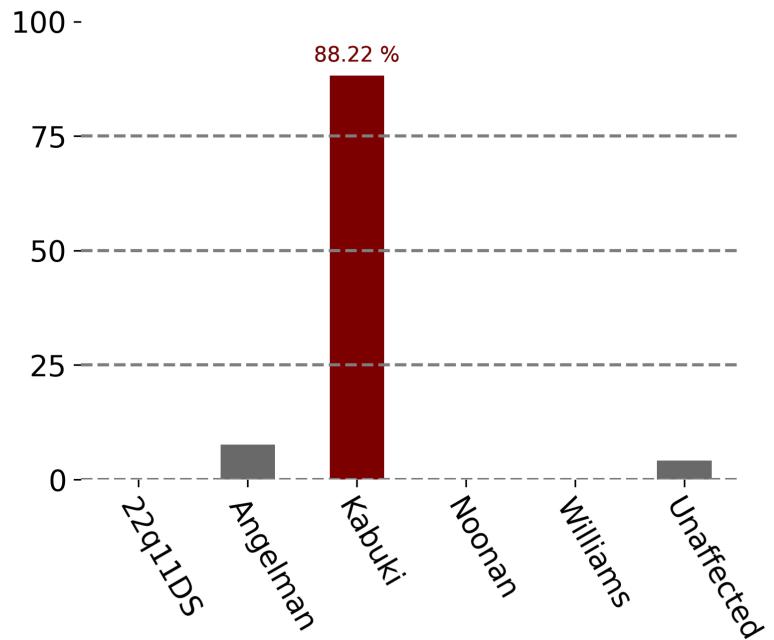
saliency map



region relevance



classifier probabilities



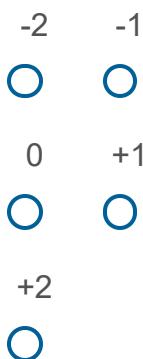
Select the genetic condition or unaffected.

Rate your confidence in the answer above. Answers can range from -2 (not confident) to +2 (highly confident).

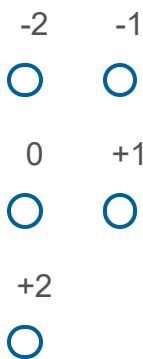


How helpful were the AI tools for this image? Answers can range from -2 (not helpful) to +2 (very helpful).

Saliency map



Region relevance



Prediction probability

-2	-1
<input type="radio"/>	<input type="radio"/>
0	+1
<input type="radio"/>	<input type="radio"/>
+2	
<input type="radio"/>	

Block 14

Please give us some feedback on the saliency maps and the region relevance scores. For your feedback you might address the following questions, but please feel free to provide any comments that come to your mind: To what extent did you consider the AI prediction when deciding whether to stay or not with your original decision? To what extent did the saliency maps and the region relevance scores influence your decision? Do you have any suggestions to us on how to improve the visual explanations? Please enter your response in the text box below.

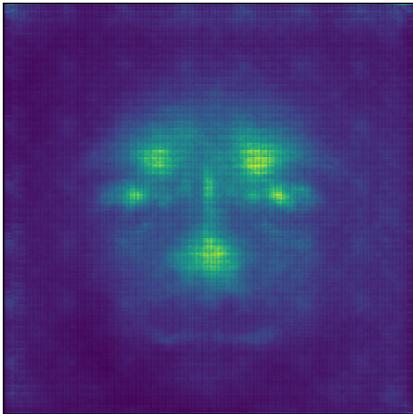
Optional

Do you have a few more minutes to look at average saliency maps to give us your thoughts?

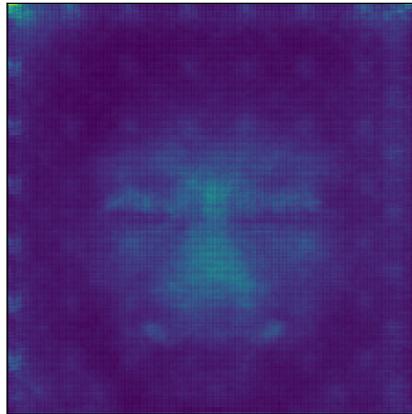
- Yes
- No

Average Saliency Maps per Syndrome (using DeepLIFT)

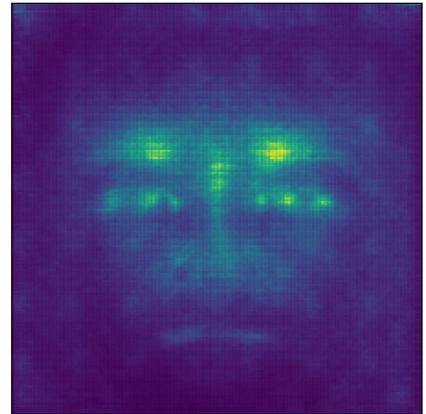
22q11DS



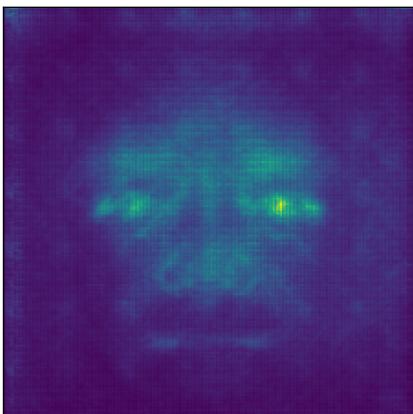
Angelman



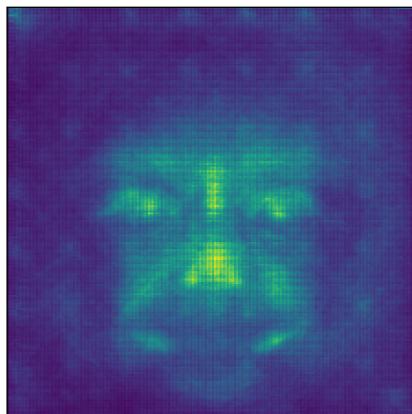
Kabuki



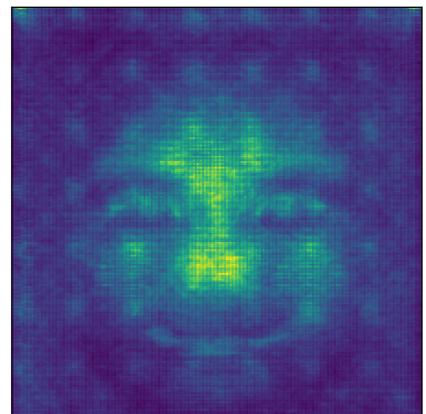
Noonan



Williams



Unaffected



Above are average saliency maps for each condition. The average saliency map is an average of hundreds of images with the same condition. The pixels the AI tool finds to be most important in classifying a genetic condition are in yellow. The brighter the area on the face, the more important the pixel is in the AI tool classification. Please take a moment to look through the images.

Which, if any, of the above average saliency maps BEST highlight the important features for that condition? Select as many or few as you like.

22q11DS

Angelman
syndrome

Kabuki
syndrome

Noonan
syndrome

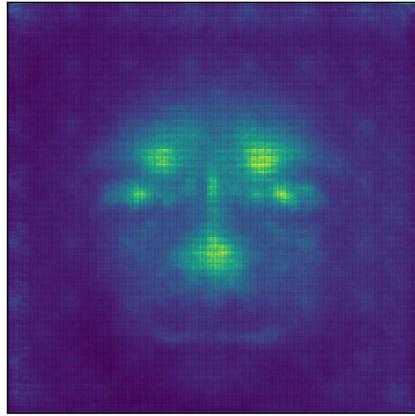
Williams
syndrome

Unaffected

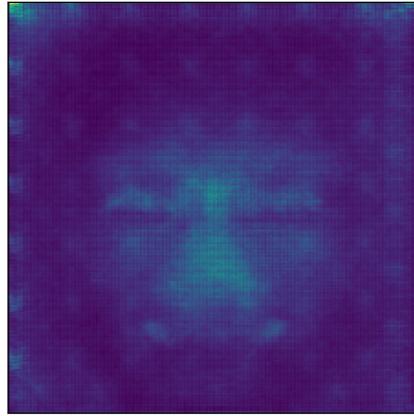
None

Average Saliency Maps per Syndrome (using DeepLIFT)

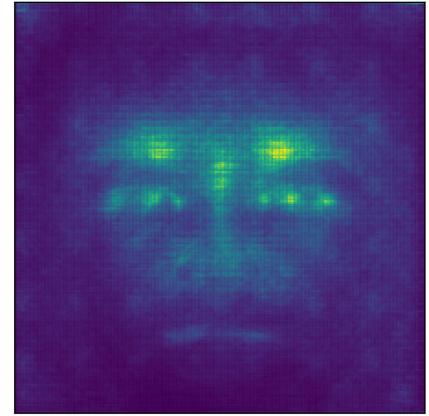
22q11DS



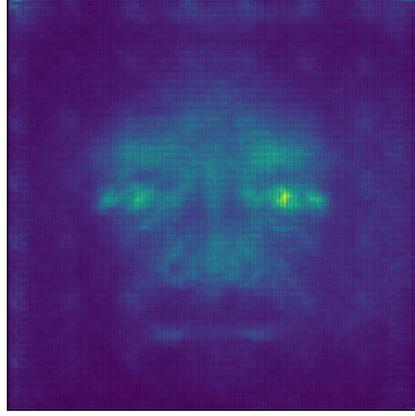
Angelman



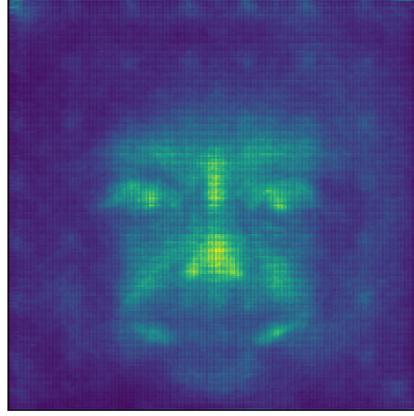
Kabuki



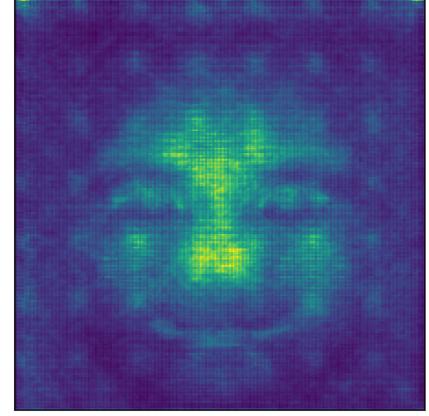
Noonan



Williams



Unaffected



What important diagnostic facial features do you see that are highlighted by the AI tool for each of the syndromes selected? Please enter your response in the box below.

Thank you

End of survey. Thank you.

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