

## Introduction and Demographic Questions

Thank you for participating in this study – we are truly grateful.

This project involves using “artificial intelligence” to help study data relevant to different genetic conditions. In this study, we are analyzing X-rays, and want to assess certain aspects by asking clinicians about the X-rays. We anticipate that participation will take most people less than 15 minutes.

We will first ask a few general questions including whether you are willing to participate and your area of expertise and experience. Next, we will show you cervical and lumbar spine x-ray images of individuals with alkaptonuria (AKU). We will ask you whether each image is from a patient receiving treatment with Nitisinone or not (no Nitisinone) treatment. We will ask for your reasoning and your overall confidence level.

You may take this survey on your smart device or your desktop/laptop computer. 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](mailto:rebekah.waikel@nih.gov) or 301.435.6558.

I am willing to participate in this study.

☐ I agree.

What is your medical specialty? Select all that apply.

- ☐ Clinical Geneticist or Medical Biochemical Geneticist (Physician)
- ☐ Radiologist

☐ Other

Please list the other medical specialty or specialties.

How many years have you been a practicing clinician (since your final residency or fellowship)?

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

## Directions

For the following questions, you will view cervical (first 10 images ) and then lumbar (next 10 images) spine X-ray images of individuals with Alkaptonuria. Some are receiving the medication Nitisinone. We will ask you whether you think the person is receiving Nitisinone, and then about any reason you may have for this choice. At the end, we will ask you about your overall confidence. 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.

## X-Rays Images

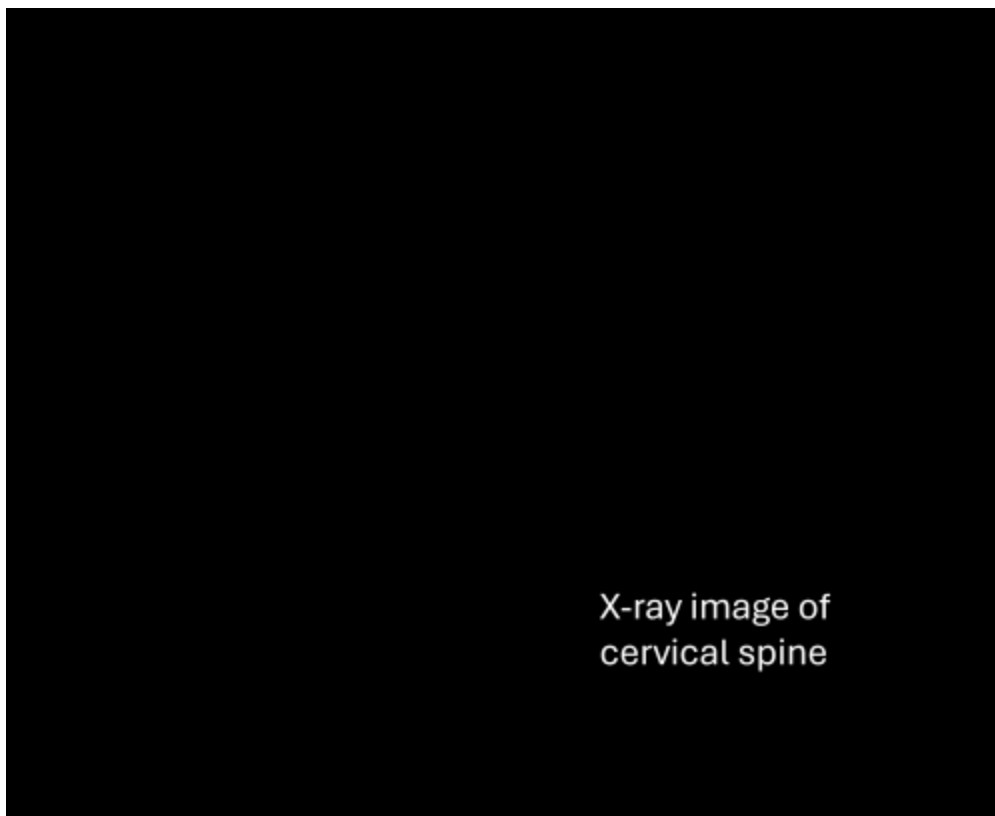
Cervical spine x-ray images



Select whether the X-ray image shows a person on therapy with Nitisinone.

- ☐ Nitisinone therapy
- ☐ No Nitisinone therapy

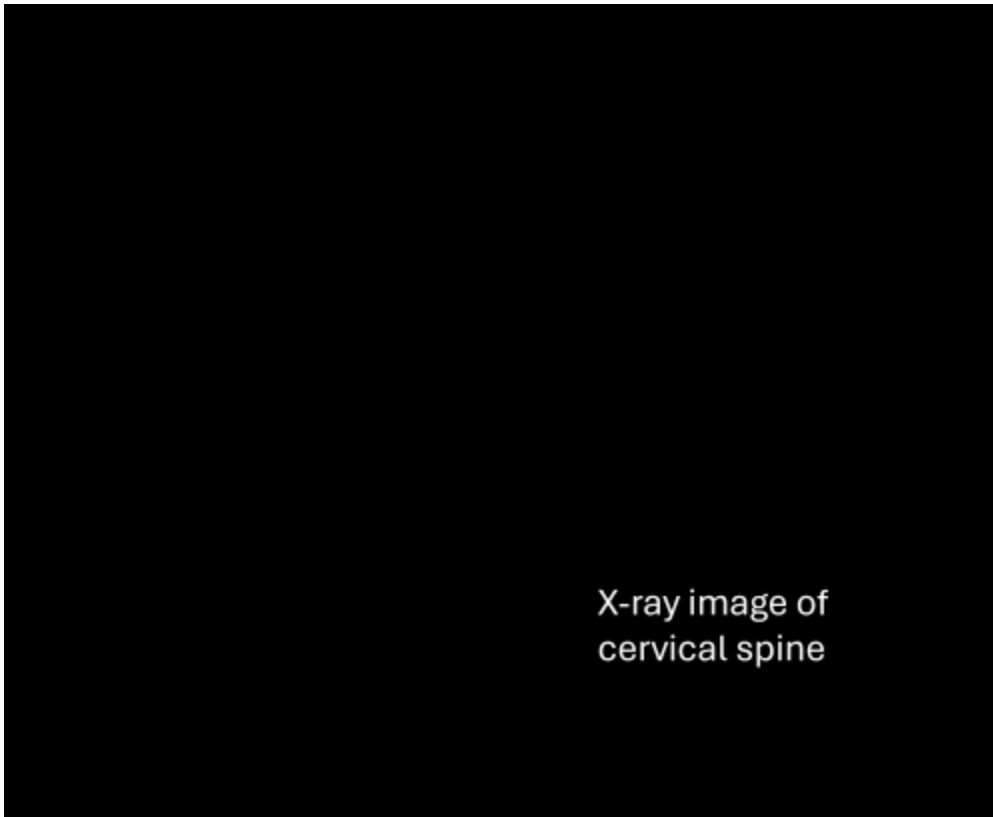
What clues in the image aided in your decision of the previous question? Enter "N/A" if no specific clues were observed or you are generally unsure why you made your choice.



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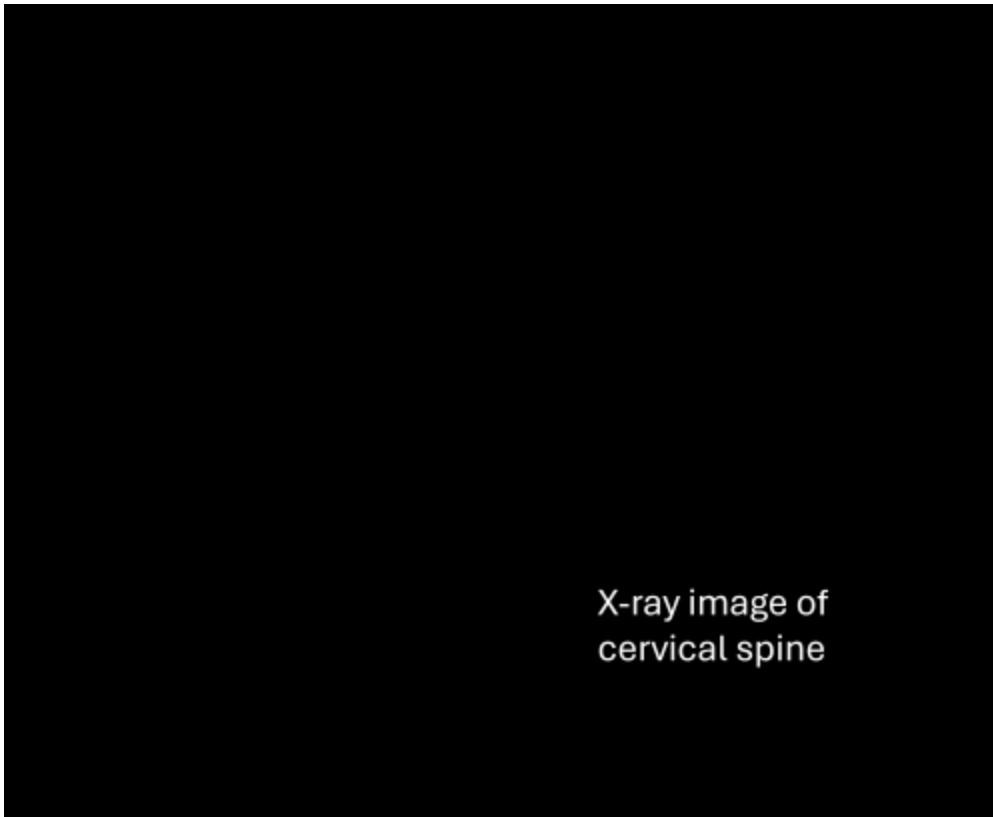
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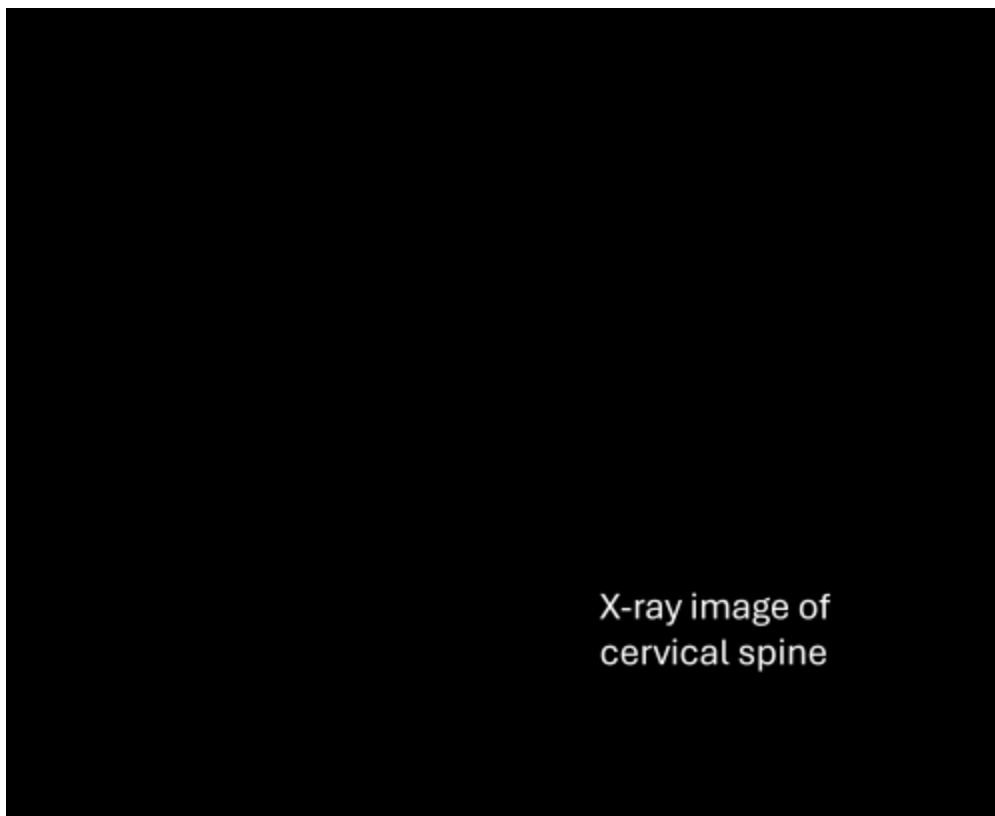
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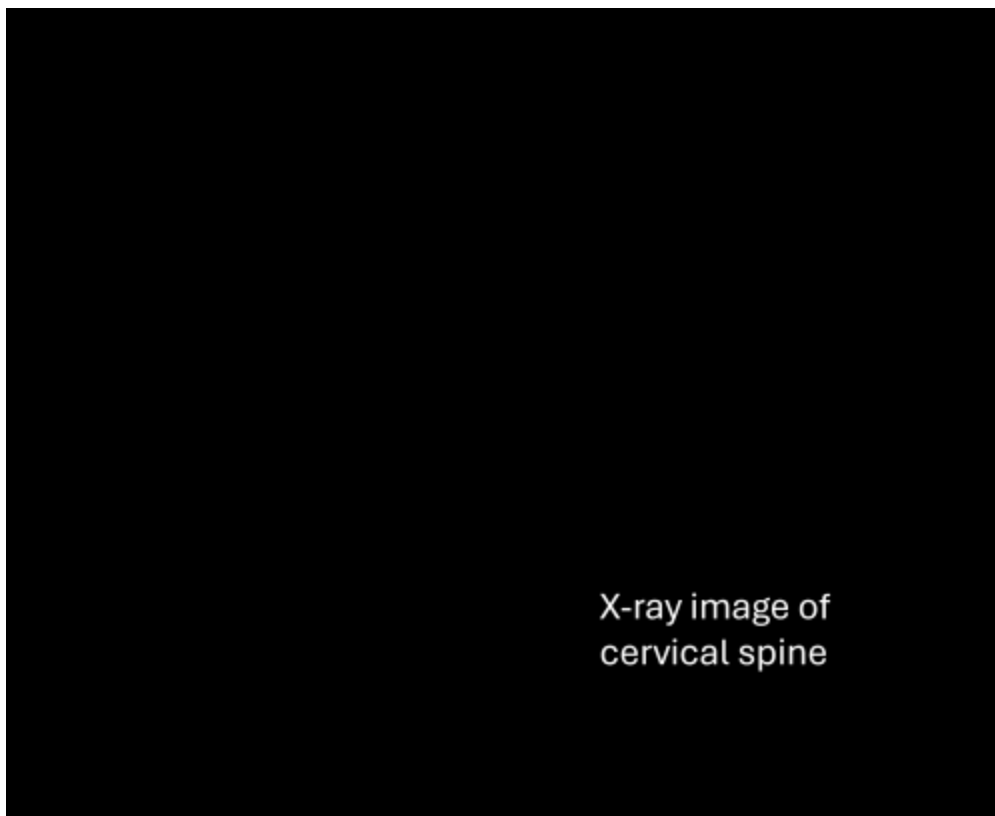
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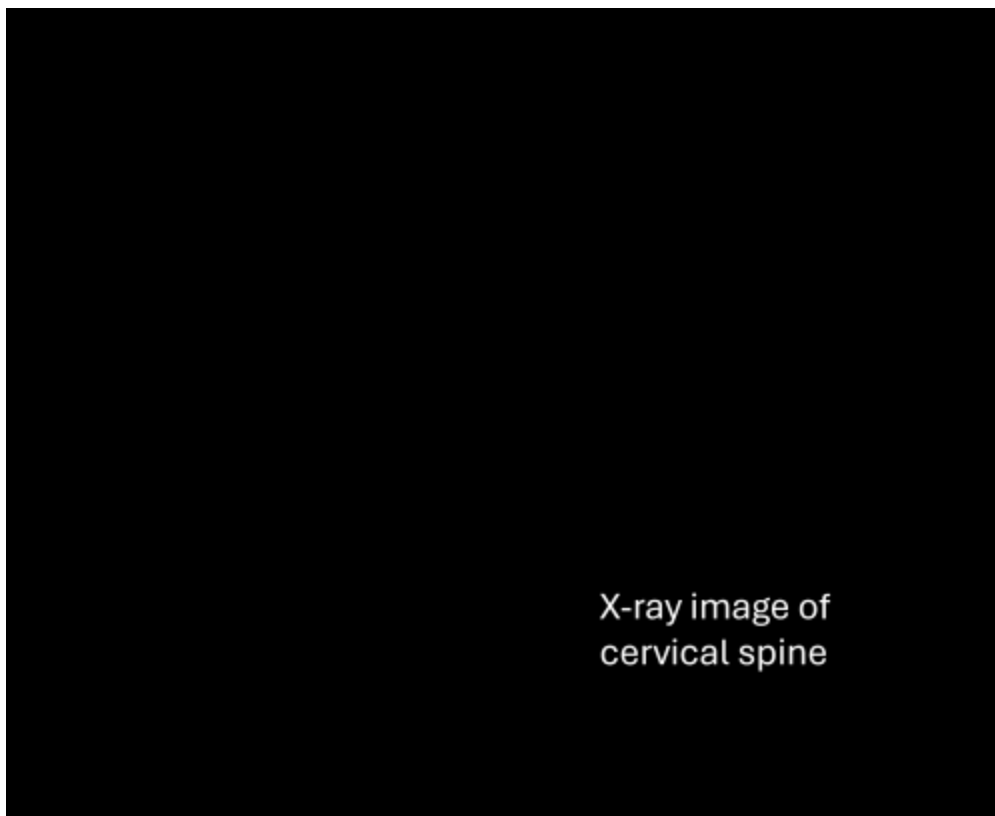


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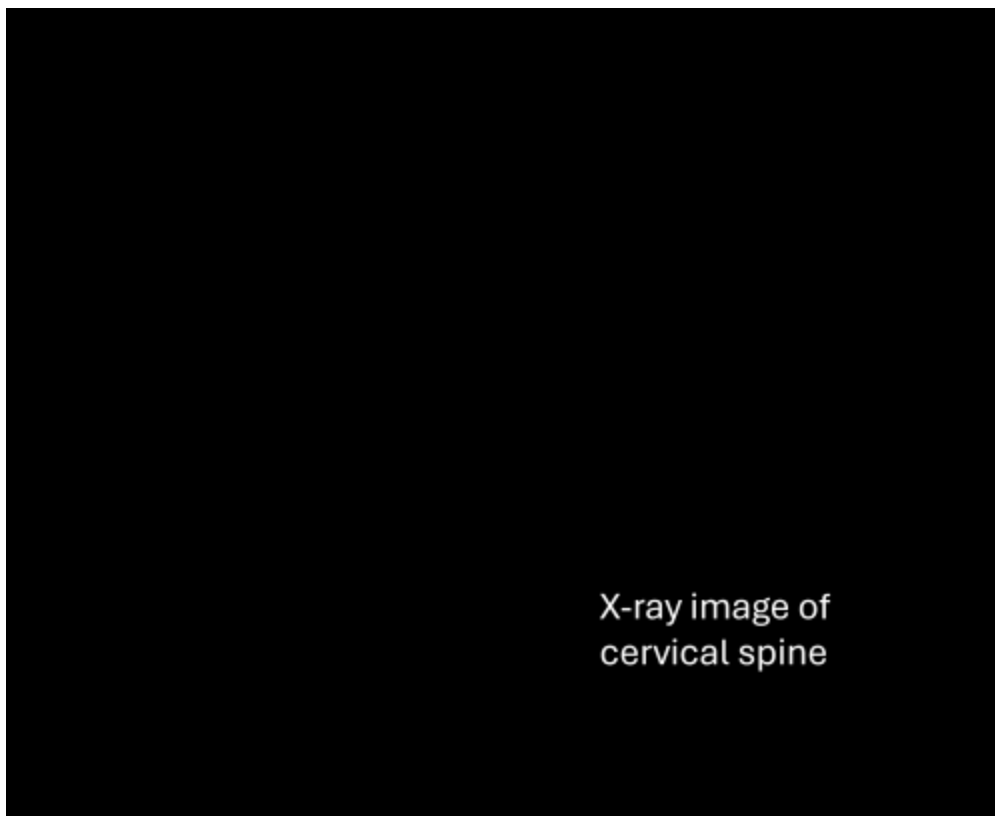




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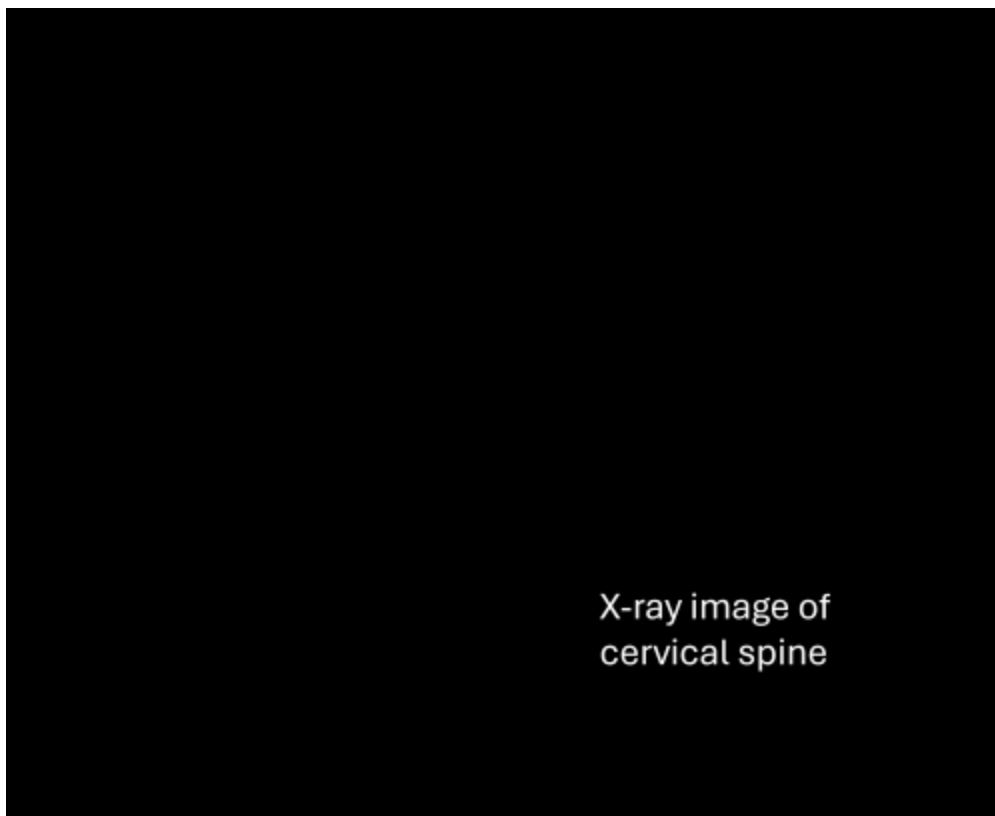


X-ray image of  
cervical spine

Select whether the X-ray image shows a person on therapy with Nitisinone.

- ☐ Nitisinone therapy
- ☐ No Nitisinone therapy

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X-ray image of  
cervical spine

Select whether the X-ray image shows a person on therapy with Nitisinone.

- ☐ Nitisinone therapy
- ☐ No Nitisinone therapy

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Select whether the X-ray image shows a person on therapy with Nitisinone.

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- ☐ No Nitisinone therapy

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Lumbar spine X-ray images



X-ray image of  
lumbar spine

- ☐ Nitisinone therapy
- ☐ No Nitisinone therapy

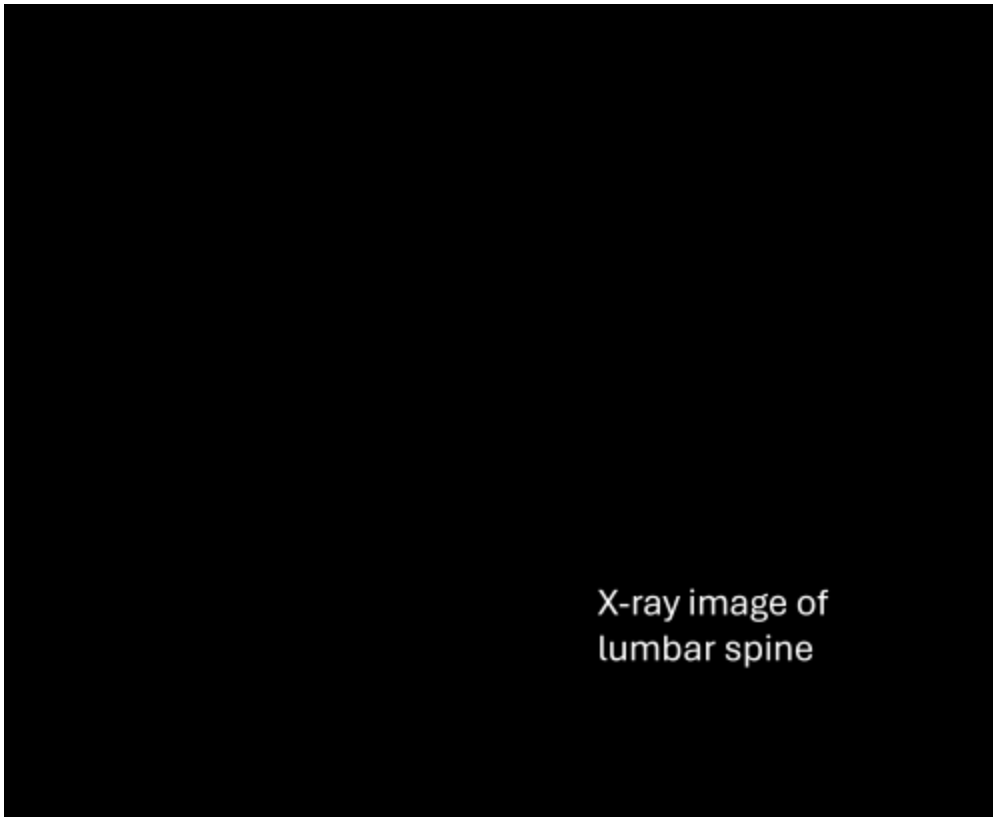
What clues in the image aided in your decision of the previous question? Enter "N/A" if no specific clues were observed or you are generally unsure why you made your choice.



X-ray image of  
lumbar spine

- ☐ Nitisinone therapy
- ☐ No Nitisinone therapy

What clues in the image aided in your decision of the previous question? Enter "N/A" if no specific clues were observed or you are generally unsure why you made your choice.

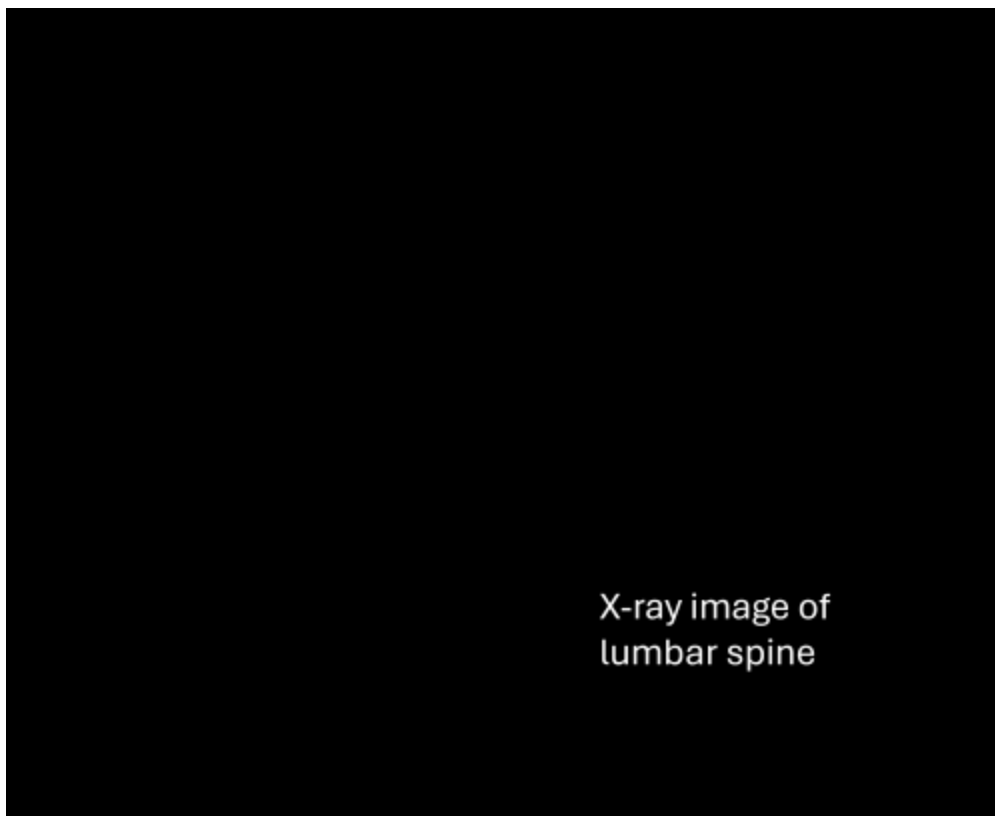


X-ray image of  
lumbar spine

Select whether the X-ray image shows a person on therapy with Nitisinone.

- ☐ Nitisinone therapy
- ☐ No Nitisinone therapy

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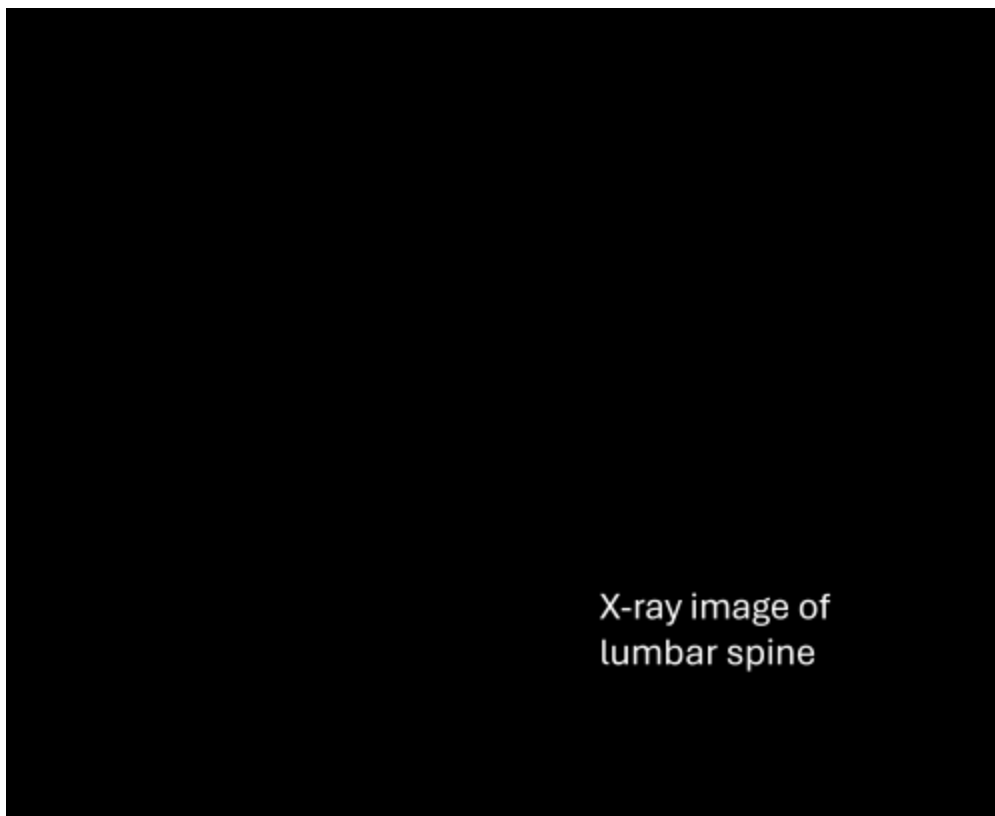
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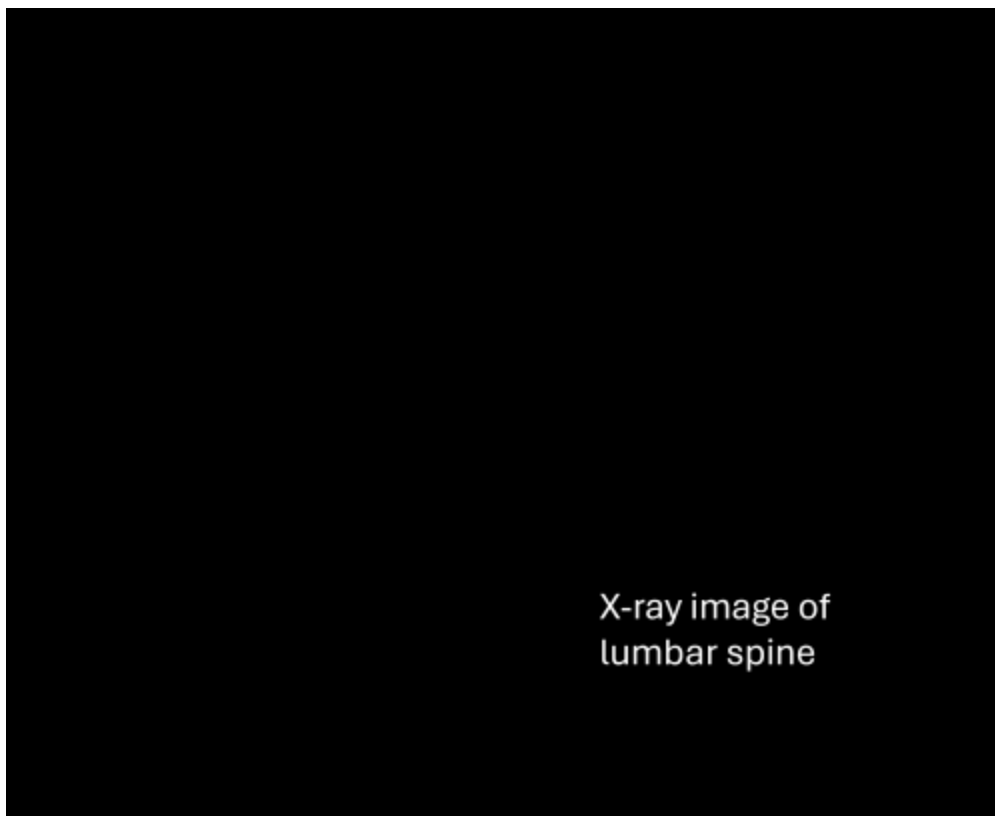


X-ray image of  
lumbar spine

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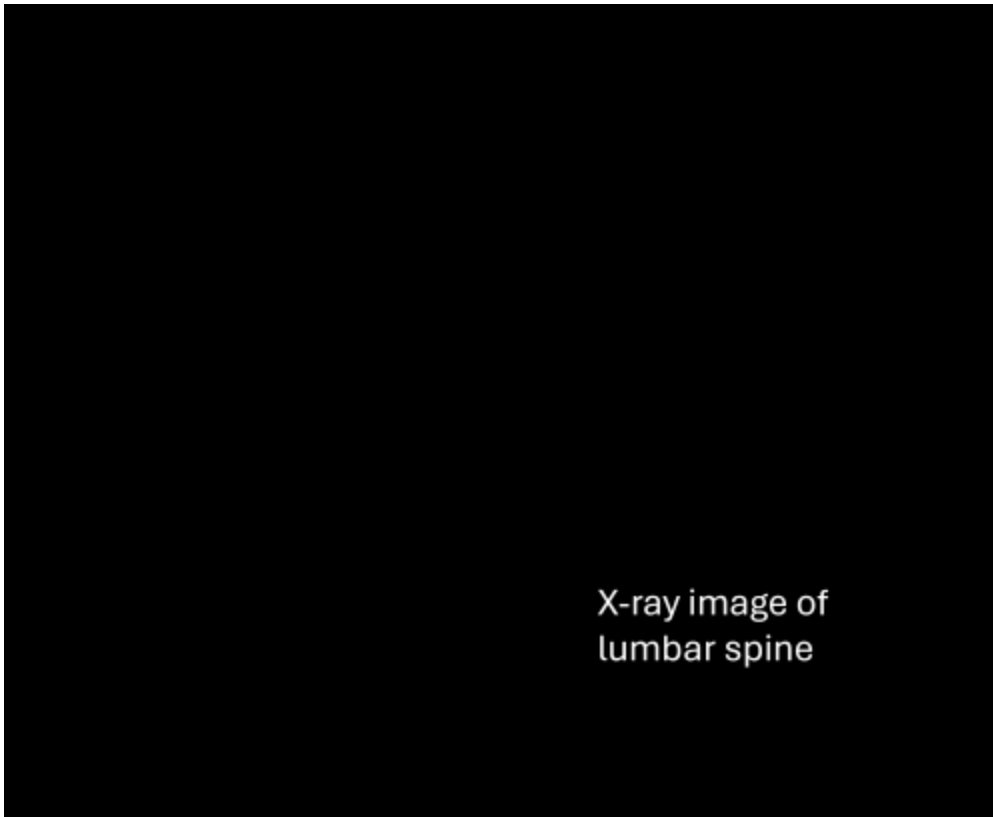
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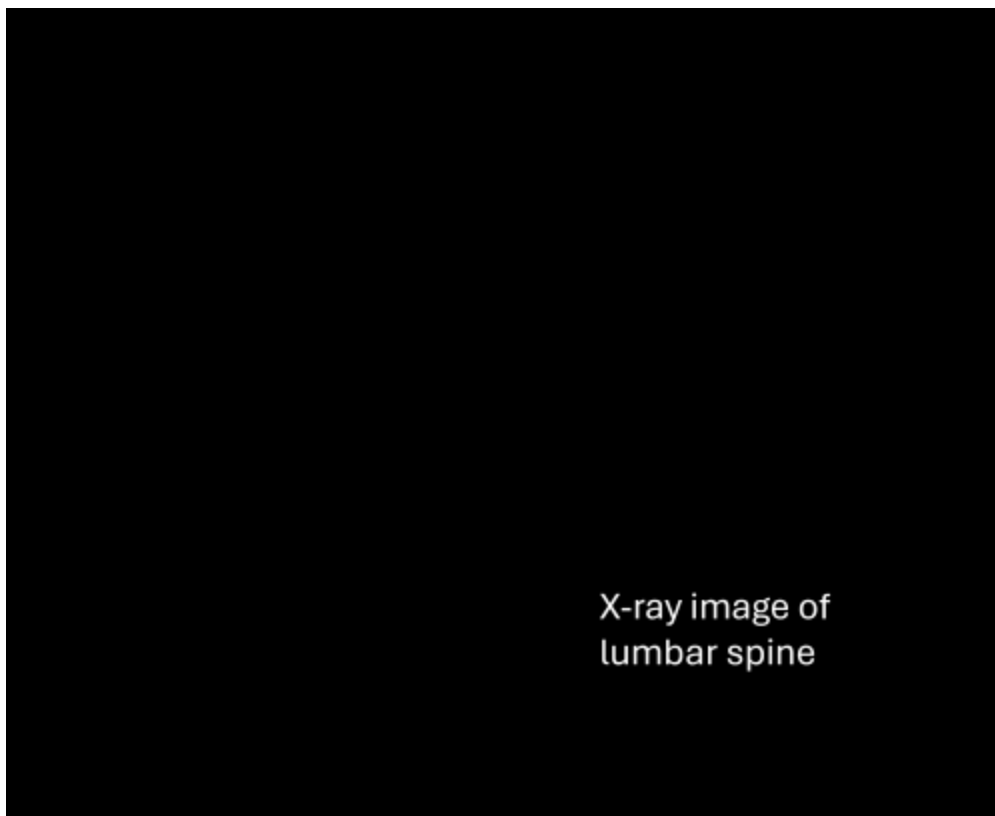
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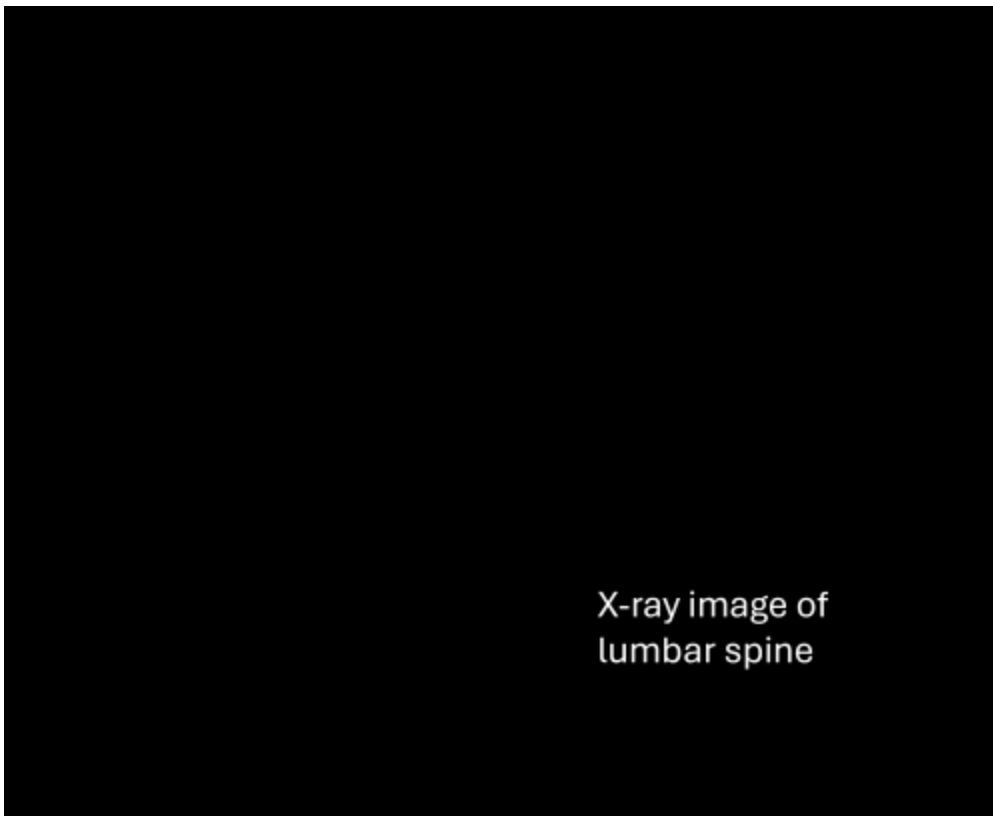


X-ray image of  
lumbar spine

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X-ray image of  
lumbar spine

Select whether the X-ray image shows a person on therapy with Nitisinone.

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- ☐ No Nitisinone therapy

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Rate your current confidence level for the following statement. I feel in general that I can identify nitsinone status in a cervical or lumbar skeletal x-ray image.

Not at all confident (0) to Highly confident (100)

0 10 20 30 40 50 60 70 80 90 100

Confidence Level

**Thank you**

End of survey. Thank you.

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