Introduction

We are grateful for your participation in this survey. This project investigates the need and the potential impact of an image recognition artificial intelligence (AI) in assisting the diagnostic process of rare bone diseases.

We anticipate that participation will take around 15 minutes.

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

Please be assured that all information you provide will be kept strictly confidential. The aggregated data may be used for research purposes, publications, or presentations, but your individual responses will remain private.

This survey is being conducted for research purposes. Your participation is voluntary. If you would like to participate, please continue. If not, you may exit from this page.

If you have questions, encounter any difficulties, or change your mind about participation, or if you are interested in further collaboration please contact Dr. Rebekah Waikel via rebekah.waikel@nih.gov.

Thank you for your time and participation.

The broader research team developing AI based tools for genetic bone conditions.
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Demographic Quesitons

Are you involved in caring for or in the diagnostic process of patients with known or possible rare bone diseases (or conditions where skeletal anomalies and related findings are an important feature)?

- ○ I am involved in the diagnostic process.
- ○ I am involved in the pre- and/or post-diagnosis patient care.
- ○ I am involved with both: diagnostic process and pre/post diagnosis care.
- ○ I am NOT involved in diagnostic process or pre/post care. Please explain your interest in this survey.

You have selected that you are not involved in either the diagnostic process or pre/post care of patients with rare

bone disease. Please describe your interest in rare bone disease.

What is your primary role in or related to healthcare?

- Physician
- Physician Assistant or Nurse Practitioner
- Genetic Counselor
- Nurse
- Researcher (but not a formal clinician)
- Other (please specify)

Briefly describe your role in healthcare.

What specialty best describes your current focus within healthcare?

- Endocrinology
- Genetics

- ○ OB/GYN
- ○ Orthopaedics
- ○ Pediatrics
- ○ Primary care (other than Pediatrics, e.g., Internal Medicine, Family or General Practice)
- ○ Radiology
- ○ Other (please specify)
- ○ Not applicable

Please enter the specialty that best describes your current clinical focus.

Which title best describes your position (please check if you have a dual function)?

- ☐ Endocrinologist
- ☐ Medical Geneticist
- ☐ Neurosurgeon
- ☐ Orthopaedic surgeon
- ☐ Pediatrician
- ☐ Radiologist
- ☐ Internist
- ☐ Family medicine physician

- [ ] Other, please specify

Please write the title or titles that best describe your position.

Are you involved in teaching or training other healthcare professionals?

- ○ Yes
- ○ No

What age group(s) of the patients do you work with (check all that apply)?

- [ ] Neonates and infants (0-1 year old)
- [ ] 1 to 10 years old
- [ ] 10 to 18 years old
- [ ] Greater than 18 years old

How many years of experience do you have in the healthcare field?

- ○ Less than 1 year
- ○ 1 to 5 years
- ○ 5 to 10 years
- ○ More than 10 years

Which type of healthcare facility best describes where you primarily work (i.e., where do you work most often)?

- ○ Academic medical center
- ○ Community hospital or clinic
- ○ Private practice
- ○ Research institution
- ○ Government medical center
- ○ Other (Please specify)

Please describe the healthcare facility where you primarily work.

Please enter the country where you primarily work.

Approximately, how many patients with known or suspected rare bone diseases (or conditions where skeletal anomalies and related findings are an important feature) do **you** see per month?

- Less than 5 per month
- 5 to 10 per month
- 10 to 20 per month
- 20 to 50 per month
- 50 to 100 per month
- 100 to 200 per month
- Greater than 200 per month

Approximately, how many patients with known or suspected rare bone diseases (or conditions where skeletal anomalies and related findings are an important feature) does **your clinic** see per month?

- Less than 5 per month
- 5 to 10 per month

- ○ 10 to 20 per month
- ○ 20 to 50 per month
- ○ 50 to 100 per month
- ○ 100 to 200 per month
- ○ Greater than 200 per month

Approximately, how many patients with known or suspected rare bone diseases (or conditions where skeletal anomalies and related findings are an important feature) does **your entire facility** see per month?

- ○ Less than 5 per month
- ○ 5 to 10 per month
- ○ 10 to 20 per month
- ○ 20 to 50 per month
- ○ 50 to 100 per month
- ○ 100 to 200 per month
- ○ Greater than 200 per month

According to the 2023 revision of the nosology of genetic skeletal disorders (Unger et al.), there are 41 different groups of skeletal disorders. Please SELECT ALL of the groups that represent the patients for which you, your clinic, and/or your institution provide care.

- [ ] FGFR3 chondrodysplasias
- [ ] Mesomelic and rhizo-mesomelic dysplasias
- [ ] Osteolysis group
- [ ] Type 2 collagen disorders
- [ ] Acromesomelic dysplasias
- [ ] Disorganized development of skeletal components group
- [ ] Type 11 collagen disorders
- [ ] Acromelic dysplasias
- [ ] Overgrowth (tall stature) syndromes and segmental overgrowth
- [ ] Sulfation disorders
- [ ] Brachydactylies (isolated)
- [ ] Genetic inflammatory or rheumatoid-like osteoarthropathies
- [ ] Dysplasias with multiple joint dislocations
- [ ] Brachydactylies as part of syndromes
- [ ] Cleidocranial dysplasia and related disorders
- [ ] Filamins and related disorders
- [ ] Bent bones dysplasia group
- [ ] Syndromes featuring craniosynostosis
- [ ] Proteoglycan core proteins disorders
- [ ] Primordial dwarfism and slender bones group
- [ ] Craniofacial Dysostoses
- [ ] TRPV4 disorders
- [ ] Lysosomal Storage Diseases with Skeletal Involvement
- [ ] Vertebral and costal dysostoses
- [ ] Pseudoachondroplasia and the multiple epiphyseal dysplasias
- [ ] Chondrodysplasia punctata (CDP) group
- [ ] Patellar dysostoses
- [ ] Skeletal disorders caused by abnormalities of cilia or ciliary signaling
- [ ] Osteopetrosis and related osteoclast disorders
- [ ] Limb hypoplasia - reduction defects group

- [ ] Metaphyseal dysplasias
- [ ] Osteosclerotic disorders
- [ ] Split hand/foot with and without other manifestations
- [ ] Spondylometaphyseal dysplasias (SMD)
- [ ] Osteogenesis Imperfecta and bone fragility group
- [ ] Polydactyly-Syndactyly-Triphalangism group
- [ ] Spondyloepi(meta)physeal dysplasias (SE(M)D)
- [ ] Disorders of bone mineralisation
- [ ] Defects in joint formation and synostoses
- [ ] Severe spondylodysplastic dysplasias
- [ ] Skeletal disorders of the parathyroid hormone signaling cascade

In your opinion, what are the **three** most challenging skeletal disorders to diagnose?

Interest in image recognition AI

In your opinion, how important are medical images (i.e., x-rays, MRI, etc.) in the diagnosis of rare bone diseases?

- ○ Not at all important
- ○ Slightly important

- ○ Moderately important
- ○ Very important
- ○ Extremely important

Which imaging type do you think is the most important modality for the postnatal diagnosis of rare bone diseases?

- ○ Projectional radiography (X-rays)
- ○ Computed tomography (CT) scans
- ○ Dual-energy X-ray absorptiometry
- ○ Magnetic resonance imaging (MRI)

How difficult do you think it is to delineate between different rare bone diseases based on visual inspection of patients' radiographs? (for answering this question you may exclude the disorders with highly characteristic features such as achondroplasia).

- ○ Extremely difficult
- ○ Somewhat difficult
- ○ Neither easy nor difficult
- ○ Somewhat easy
- ○ Extremely easy

Optional: Please add any thoughts/comments you have about ease or difficulty delineating rare bone diseases by radiographs.

If an image recognition AI is developed that provides you with a prioritized list of syndromes based on a radiograph, how likely are you to consider integrating it into your current diagnostic workflow?

- Extremely unlikely
- Somewhat unlikely
- Neither likely nor unlikely
- Somewhat likely
- Extremely likely

Have you previously used or are you currently using any digital tools (computer software, AI, or machine learning) in your diagnostic workflow?

- Yes, for image analysis. Please name the tool or the task on the next page.

- ○ Yes, for data interpretation (e.g., genomics data). Please name the tool(s) on next page.
- ○ Yes, for other tasks. Please name the task(s) on next page.
- ○ No, but considering implementation. Please name the tasks on next page.
- ○ No, and not considering it.

What tools and or tasks did you use AI based image analysis?

What tools did you use for data interpretation?

Please describe the tasks.

Please describe the tasks.

Are there specific areas (other than image recognition) within the diagnostic, management, or treatment process where you believe AI could make a positive impact?

Would you be willing to do additional training to learn to utilize an image recognition AI tool for rare bone disease diagnosis?

- Yes
- No
- Unsure

Concerns and Reservations

What regulatory considerations or ethical concerns do you foresee in implementing image recognition AI for rare bone disease diagnosis? Select all that apply.

- [ ] Data privacy and security
- [ ] Interpretability of AI algorithms
- [ ] Compliance with medical regulations
- [ ] Informed consent for AI-based diagnostics
- [ ] Other (please specify)

Please describe other concerns in implementing image recognition AI for rare bone disease diagnosis.

How concerned are you about the potential for AI-related errors in the diagnosis of rare bone diseases?

- ○ Very concerned
- ○ Somewhat concerned
- ○ Neutral
- ○ Not very concerned
- ○ Not concerned at all
- ○ Unsure, need to see research results first.

As long as the image recognition AI algorithms are confirmed (through trial studies) to perform their tasks

accurately, how important to you is the explainability of these algorithms?

- ○ Not at all important
- ○ Slightly important
- ○ Moderately important
- ○ Very important
- ○ Extremely important

Optional: Please provide any additional feedback about this survey or AI based tools in diagnosis of rare bone disease.

Optional: If you like us to contact you for further collaboration, please enter your email address below.

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