



DEPARTMENT OF HEALTH & HUMAN SERVICES

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Our research group at the National Human Genome Research Institute (NHGRI), part of the National Institutes of Health (NIH), develops and studies computer-based technologies to better understand medical conditions that have known or suspected genetic causes. Our goals are to better understand how these conditions develop, what causes them, and how to help with diagnosis and management.

One of our studies involves studying how different artificial intelligence (AI) models can identify genetic conditions. We are comparing how well different models work. We think this is important since these models may be a growing part of healthcare.

For this study, we are trying to collect short descriptions from people who have genetic conditions so that we can test these models. If you or your child has a genetic condition, we are hoping you could send us **one paragraph describing the condition** in you or your child. Please use whatever description is most natural for you. That is, use your own words in the way you might describe the condition to a clinician meeting you for the first time.

We will analyze these de-identified descriptions with different tools. We will compare these tools, including public models such as ChatGPT or Bard. Once we share this information with the public models, they may share or use it again. You can send these descriptions to our group through the following emailing address: medicalgenomicsunit@nih.gov

Here are some short example descriptions:

Condition: Familial Mediterranean Fever

My daughter is fifteen years old. She started getting attacks of fever, red rash on her feet and ankles, joint pain, and belly pain when she was about 8 years old. The attacks usually last about 1 or 2 days, and happen every few weeks, but sometimes don't happen so often. Nothing seems to trigger these attacks. She's otherwise healthy, and nobody else in the family has this.

Condition: Van der Woude syndrome

I am a 24 year-old man. I was born with a cleft lip and palate and round pits on my lower lip. I had to have surgery for the cleft lip and palate. My mother also had a cleft lip and palate.

Thank you very much. You can contact us at the addresses below if you have any questions.

Sincerely,

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