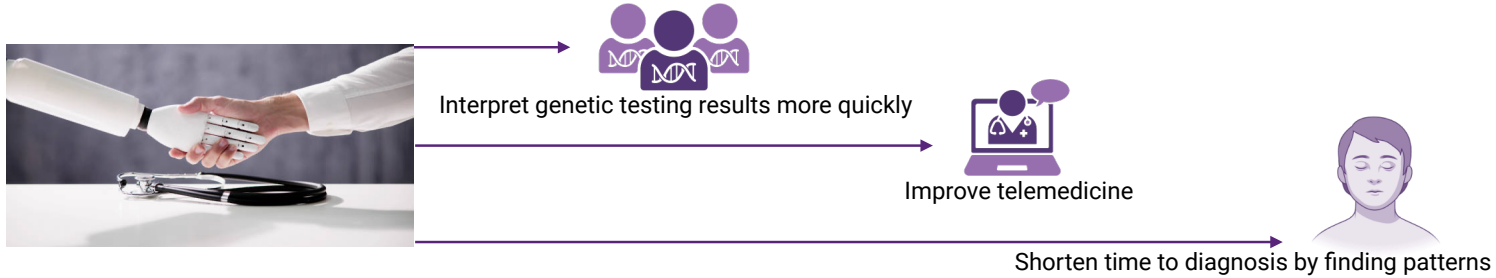


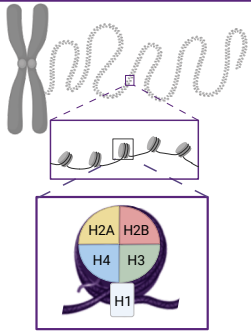
# Artificial intelligence (AI) supporting clinical and research efforts for ultra-rare disorders

## How is AI being integrated into clinical genetics?

- A recent special issue of the scientific journal *The American Journal of Medical Genetics* explored how clinicians and AI are partnering to:



## Leveraging AI for histone-based syndromes



### Histone-based syndromes

- H1 - HIST1H1E syndrome
- H3.3 - Bryant-Li-Bhoj Syndrome
- H4 - Tessadori-Bicknell-van Haaften Neurodevelopmental Disorder

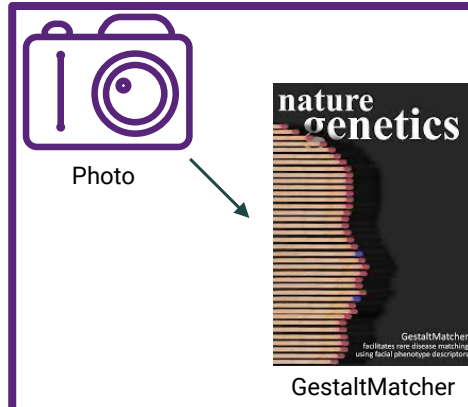
Two questions our research and clinical community would love to answer are:

- Do syndromes caused by germline variants in histones have a characteristic pattern of facial features?
  - We are asking this as we work towards shortening the time to correct diagnosis for families.
- Do all histone-based syndromes have the same pattern of facial features or do patterns of facial features vary depending on the unique syndrome?
  - We are asking this to determine if different research groups' work can apply to the whole community.

To start answering those questions, we are currently working on a study where we import published using photos of individuals with histone-based syndromes into an AI facial analysis program called GestaltMatcher to explore patterns.

The developers of GestaltMatcher put together a few overview videos available on YouTube. If you are interested in hearing them describe their platform, we have included the links below:

- <https://www.youtube.com/watch?v=pEIOaL123tY&t=17s>
- <https://www.youtube.com/watch?v=IEF7pmVOs9c>
- <https://www.youtube.com/watch?v=e2wtyOfE-Eg>



## Why are we sharing this information?

1. We think it is important to keep families updated on the research questions we are asking.
2. Families have been excited to hear about AI technology being employed in a helpful way, and how clinicians and scientists are integrating these tools into practice.
3. In the future, there may be opportunities for families to directly participate in projects like this one, and we want everyone to feel comfortable asking questions. Patient and family consent, as well as patient privacy are of the utmost importance. Before a family ever participates in a project like this, they would first discuss the risks and benefits with an enrollment coordinator who would walk through the consent process first.