

# Please join us for the HIST<sub>1</sub>H<sub>1</sub>E Syndrome Community Panel

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- Who:** Members of the Histone Team from the lab of Dr. Elizabeth Bhoj, MD, PhD at the Children's Hospital of Philadelphia
- What:** Our team is passionate about advancing research focused on histone-based genetic syndromes. We are excited to introduce ourselves and the work we are doing! We hope that this is the first of many conversations with the HIST<sub>1</sub>H<sub>1</sub>E Syndrome Community so that our work can continue to be motivated by the factors most important to you and your families.
- When:** Friday, May 17, 2024 at 12pm Eastern Time/9am Pacific Time
- Where:** <https://pennmedicine.zoom.us/j/97275039328>  
\*For privacy, this meeting will not be recorded but we welcome you to reach out!

# HIST1H1E Syndrome Community Panel

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## Agenda



Introductions



What's new in Histones



Questions



Community Considerations



**BHOJLAB**

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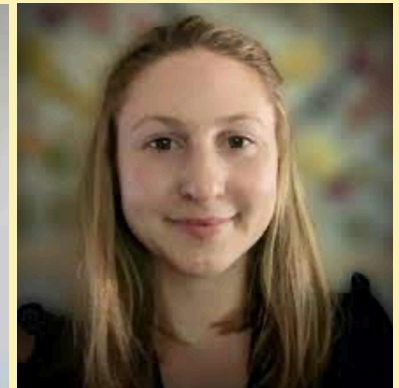
Community Considerations



**BHOJLAB**

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Introductions



**What's new in Histones**



Questions



Community Considerations



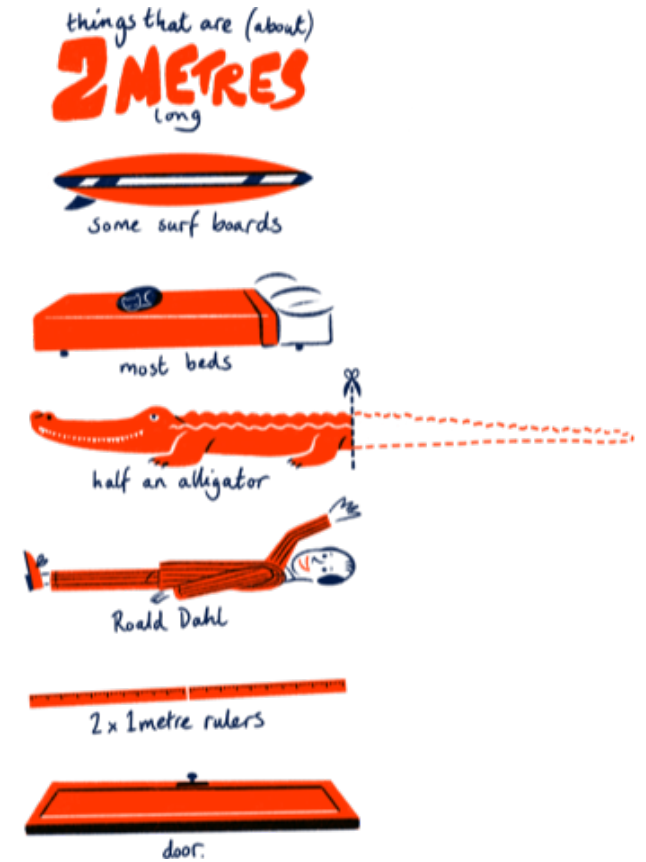
**BHOJLAB**

# Histones

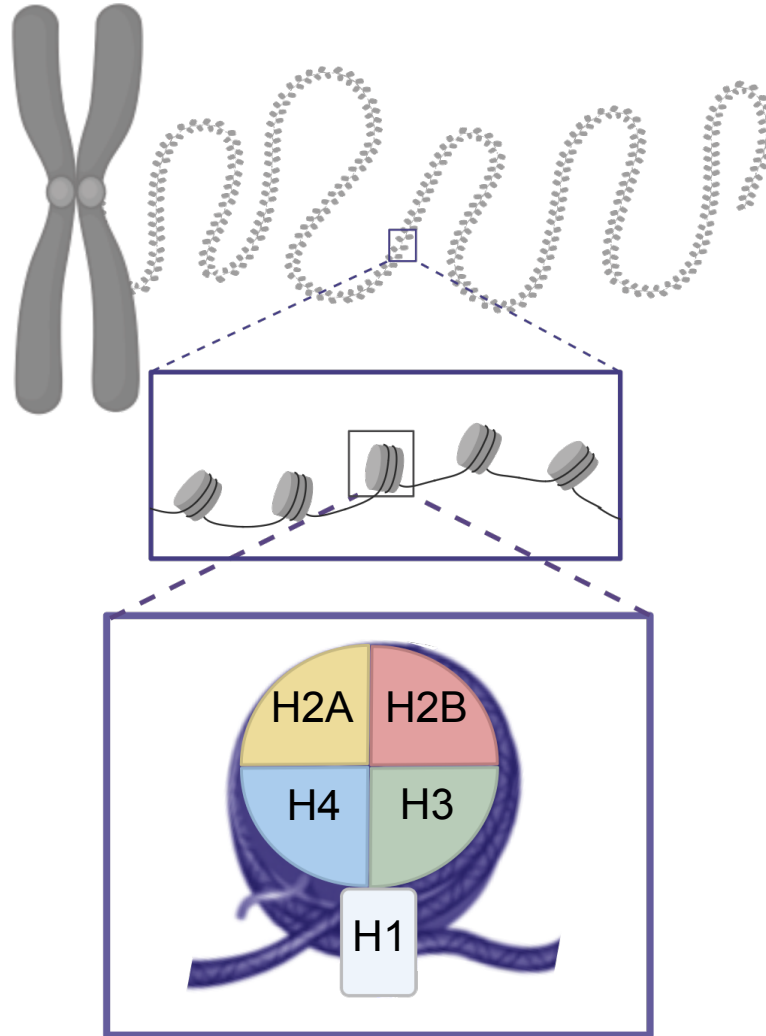
ABOUT  
**TEN**  
THOUSAND  
OF THE  
CELLS IN  
YOUR BODY  
COULD FIT  
ON THE HEAD  
OF A PIN.



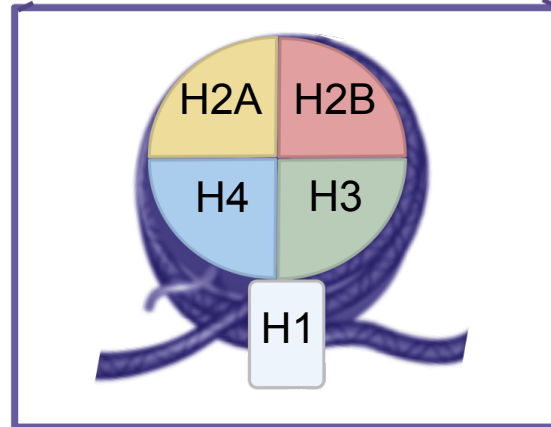
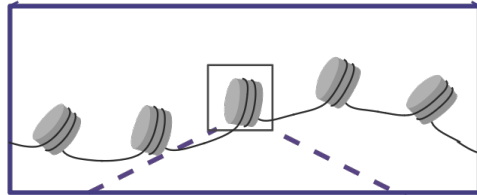
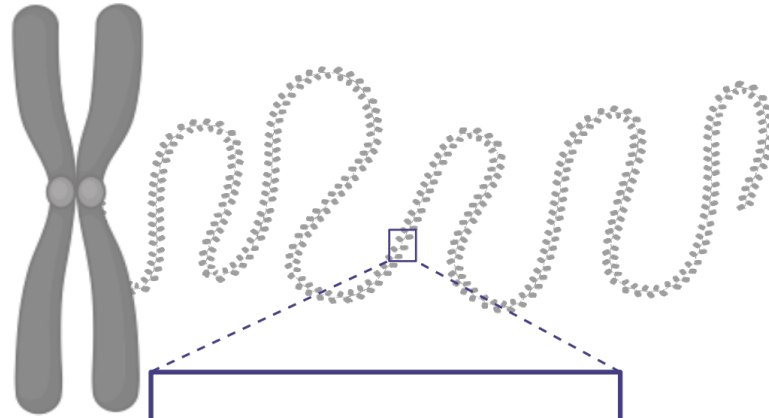
# Histones



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# Histone Syndromes



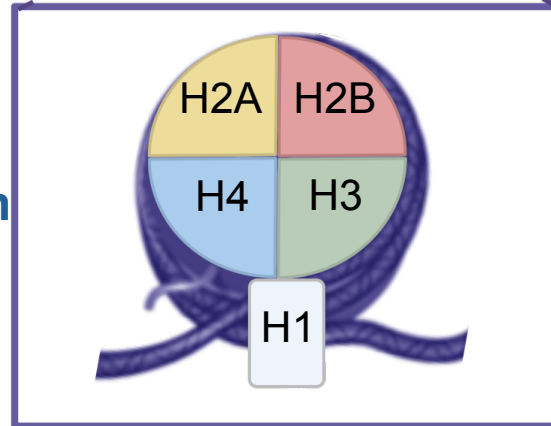
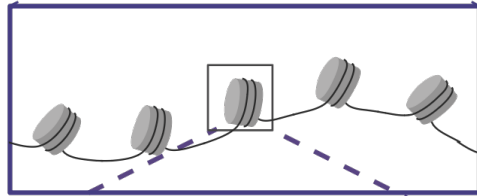
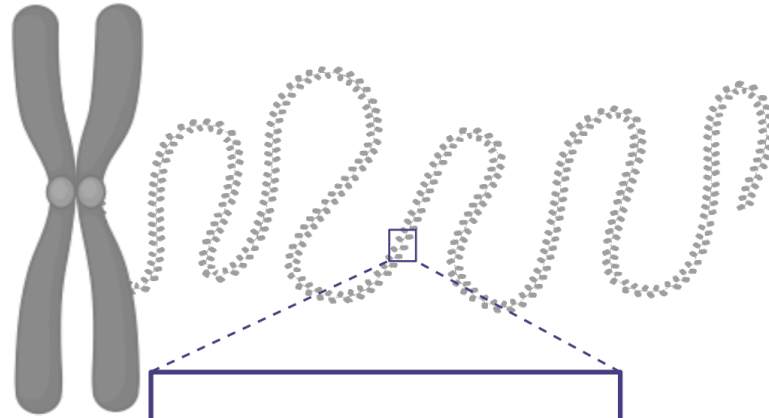
Tessadori-Bicknell-van Haaften  
Neurodevelopmental Disorder  
34 published individuals

**Bryant-Li-Bhoj Syndrome**  
**96 published individuals**

HIST1H1E Syndrome  
55 published individuals



# Histone Syndromes

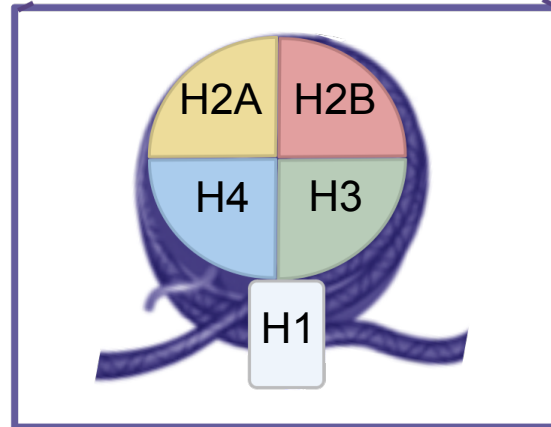
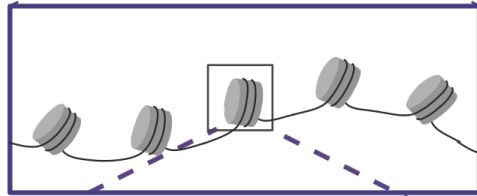
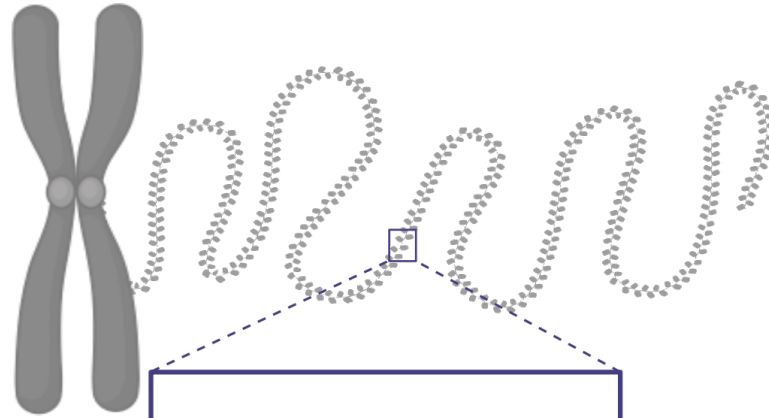


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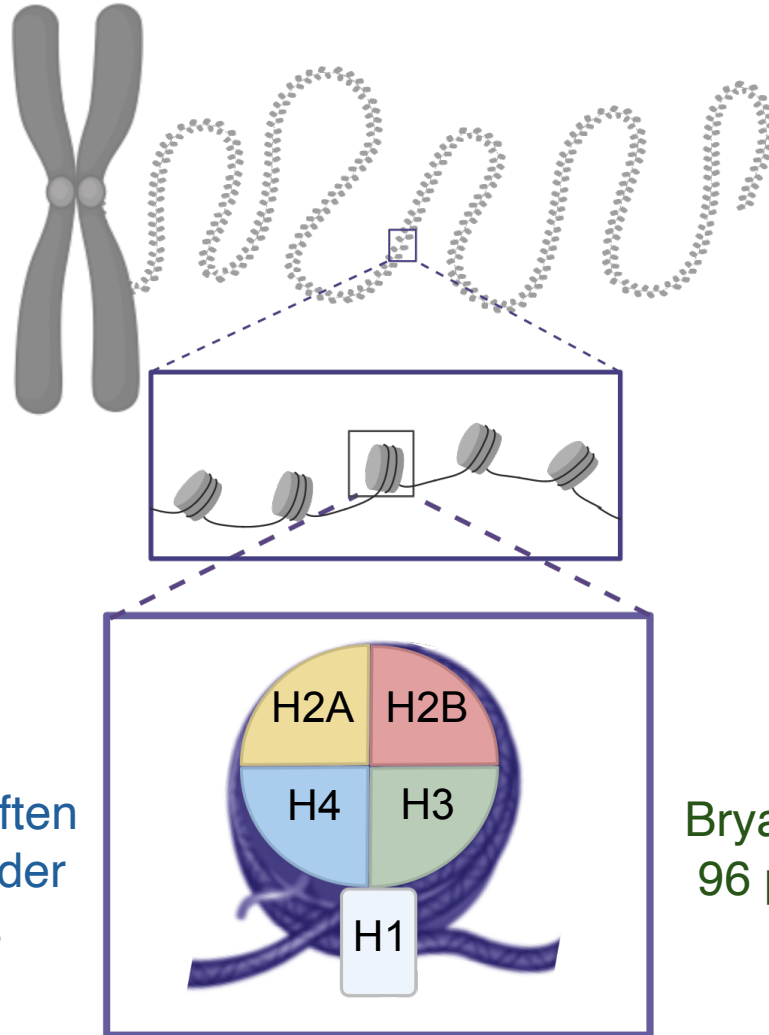


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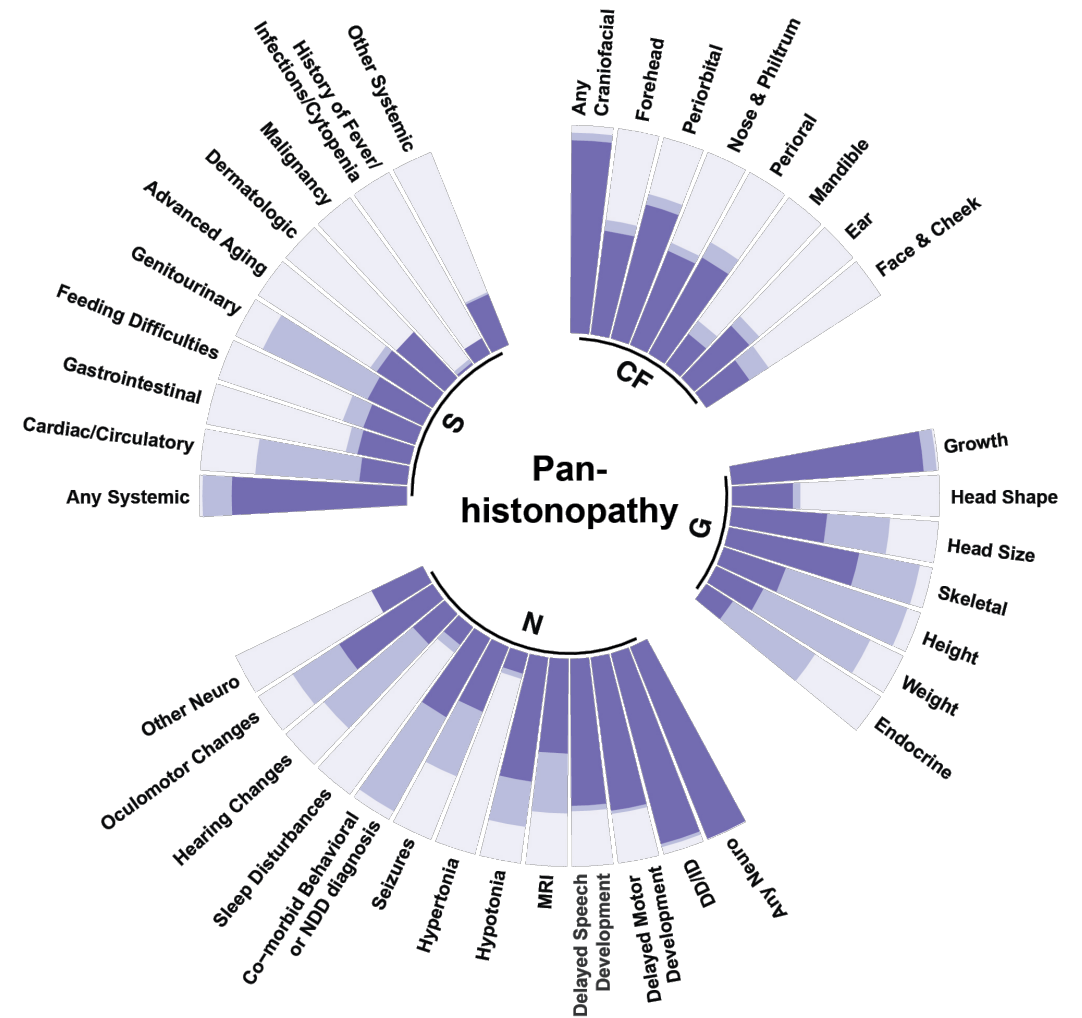
HIST1H1E Syndrome  
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Bryant-Li-Bhoj Syndrome  
96 published individuals

Previously  
unpublished  
individuals:

- HIST1H2AL/  
H2AC16
- H2AFZ/H2AZ1
- HIST1H3D/H3C4
- HIST3H3/H3-4
- 2 additional H3F3B/  
H3-3B

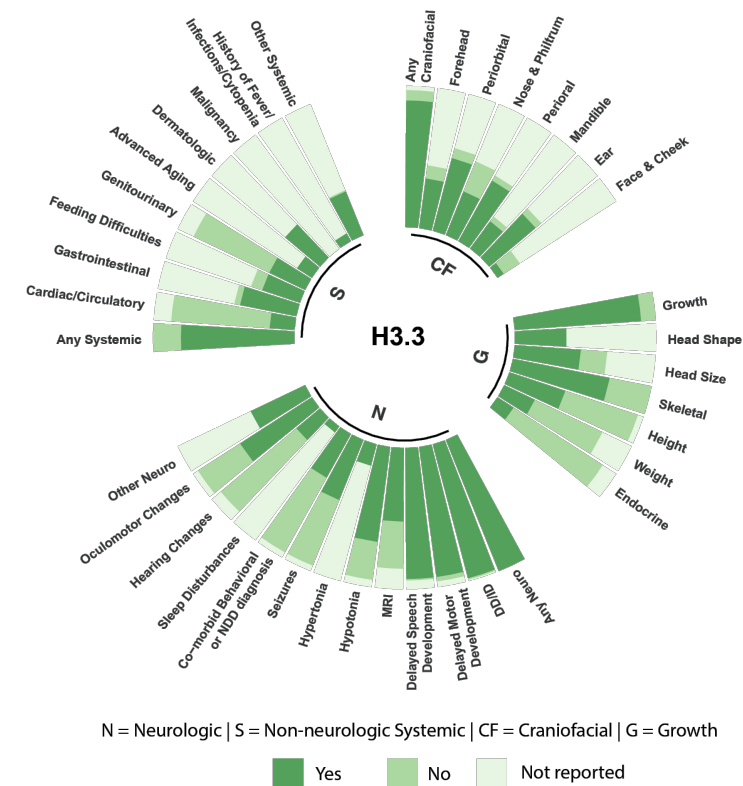
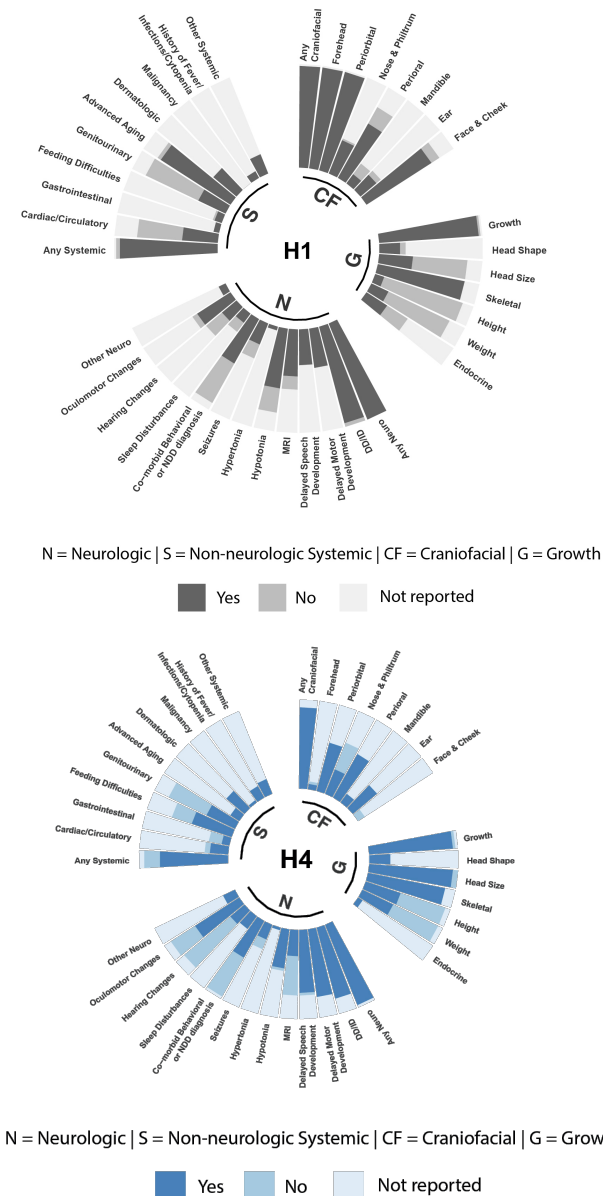
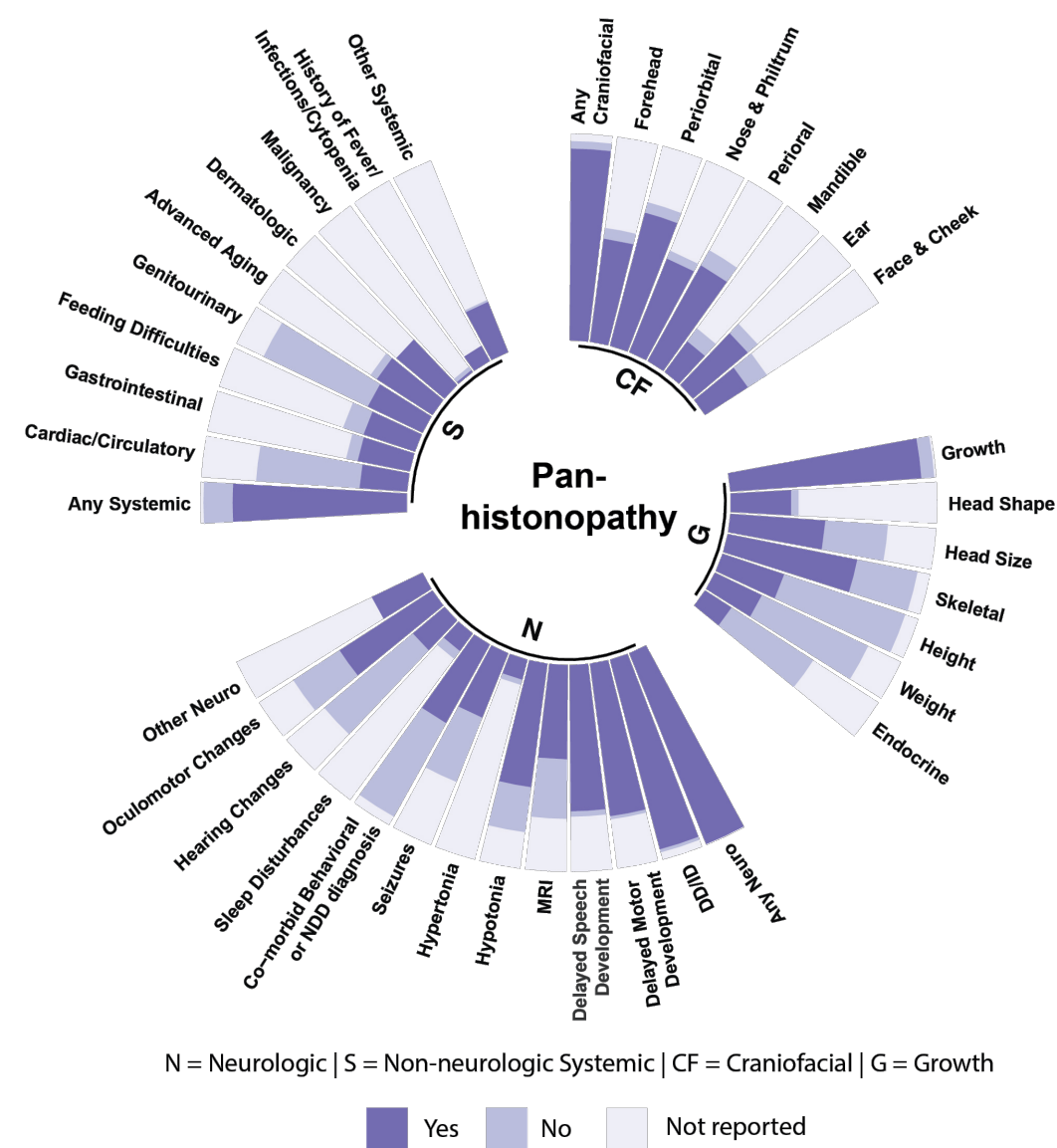
# Analyzing Clinician-Reported Features



N = Neurologic | S = Non-neurologic Systemic | CF = Craniofacial | G = Growth

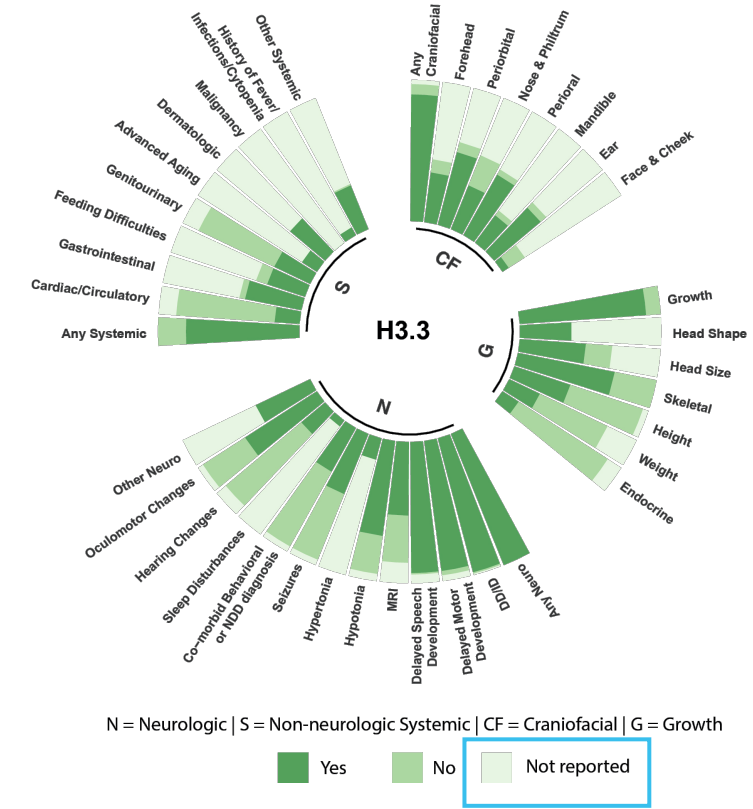
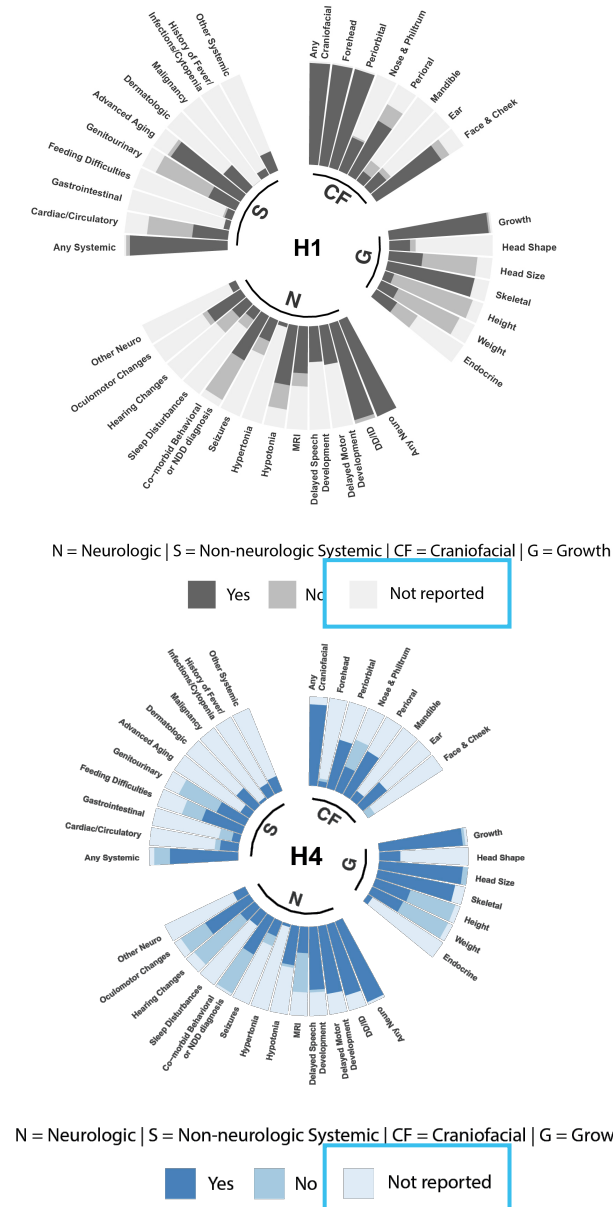
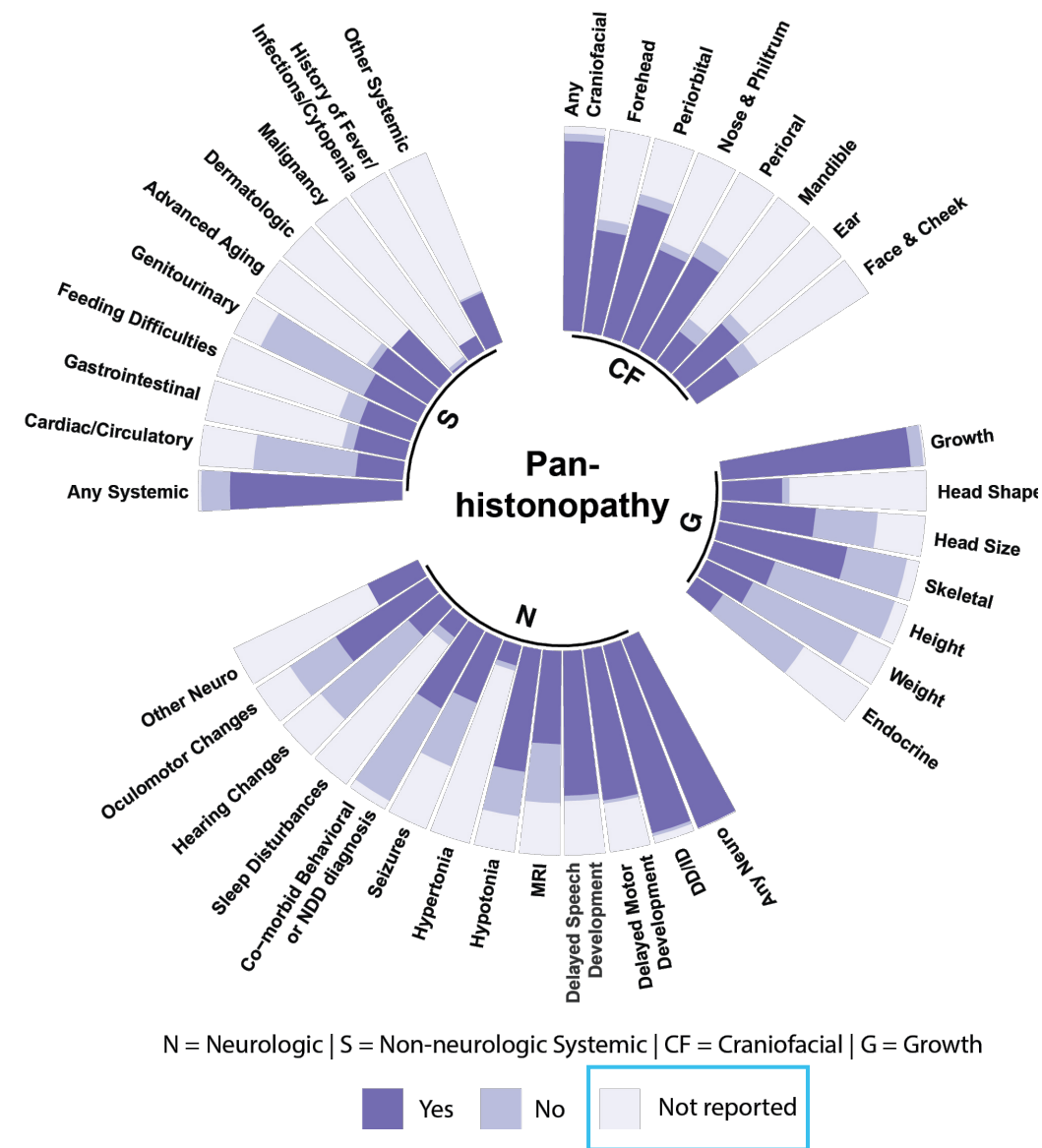
Yes No Not reported

# Analyzing Clinician-Reported Features

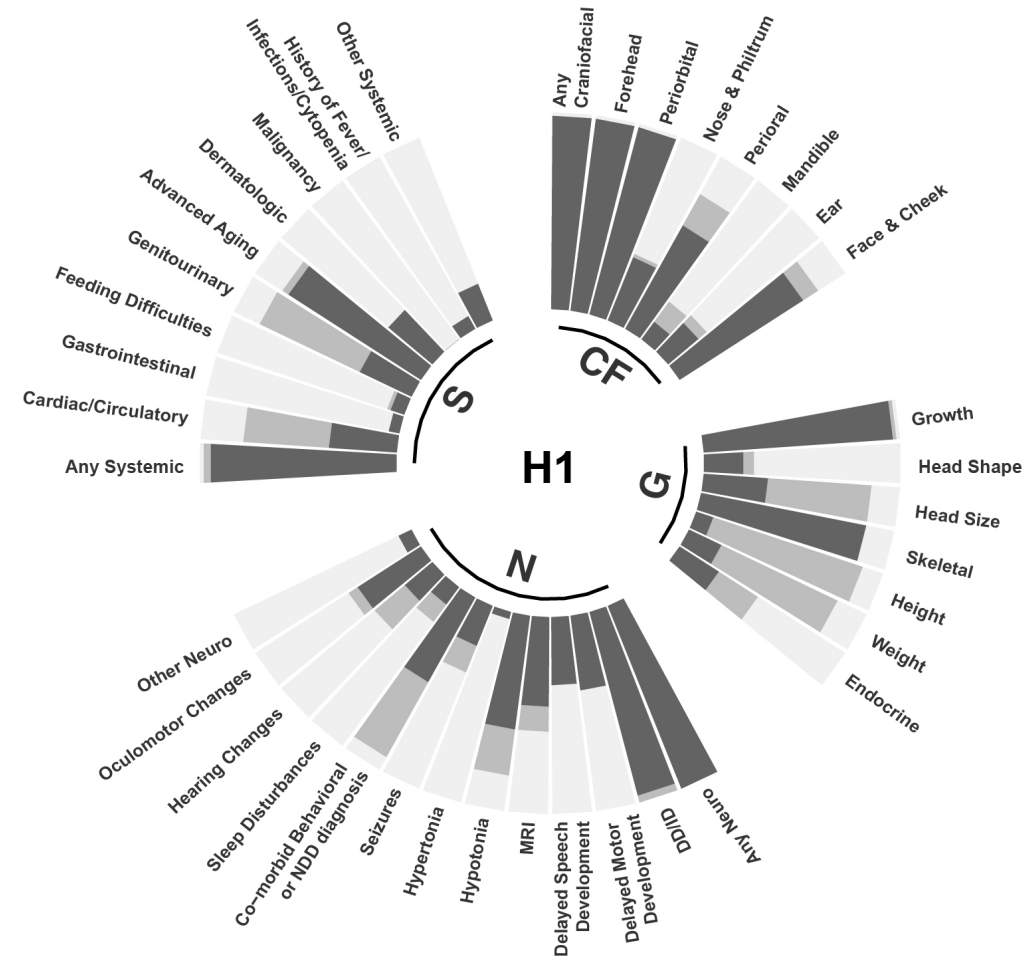




# Histone Syndromes – Patterns of Features



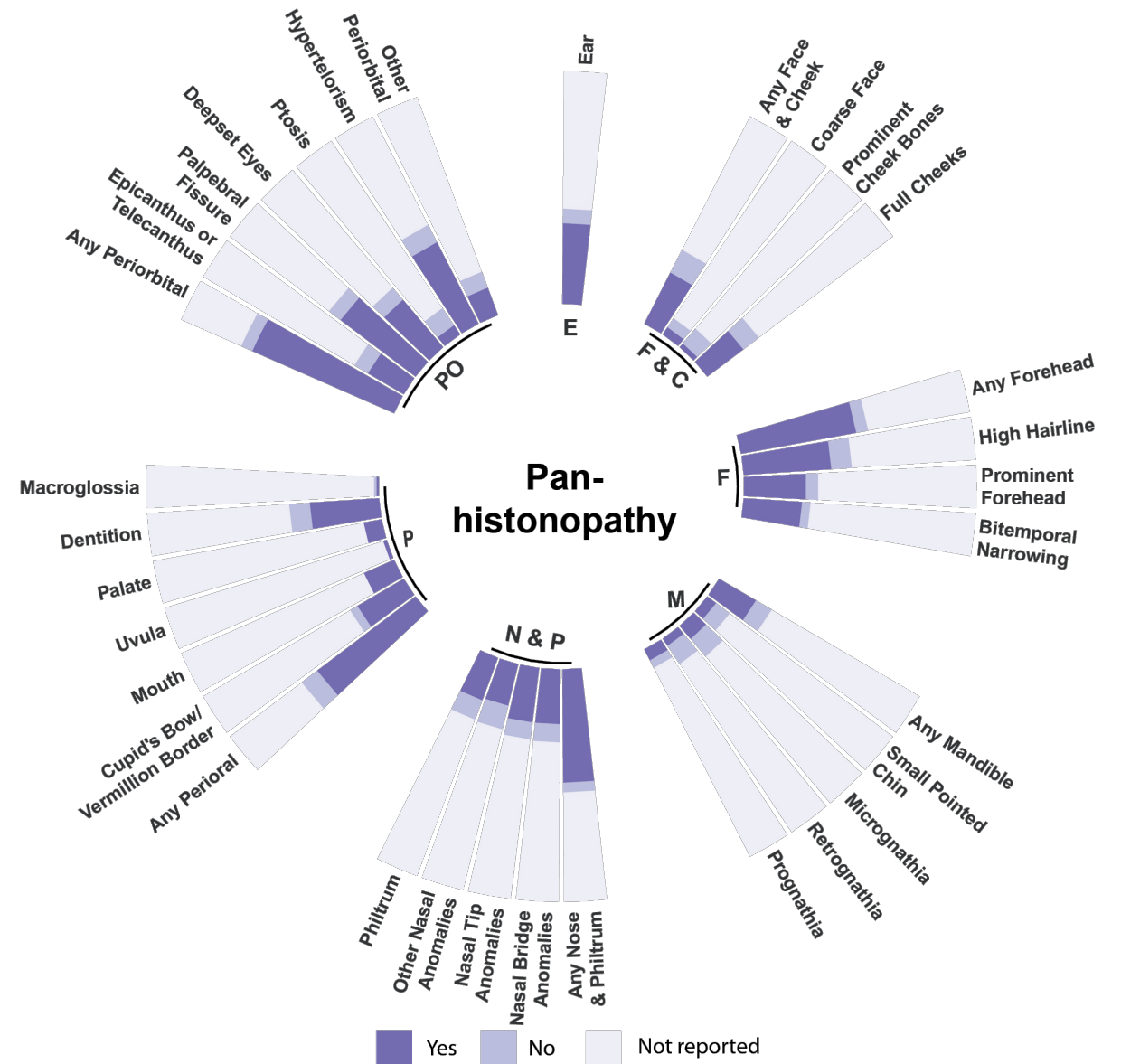
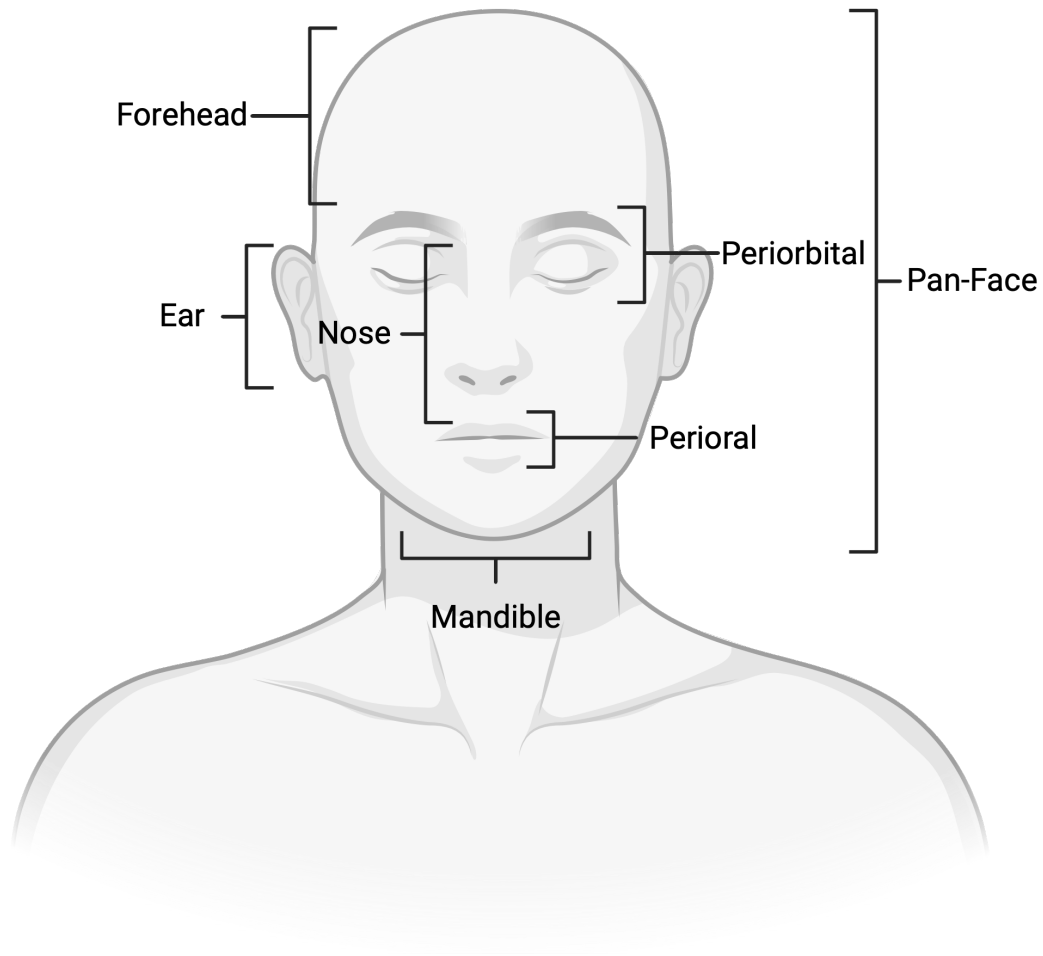
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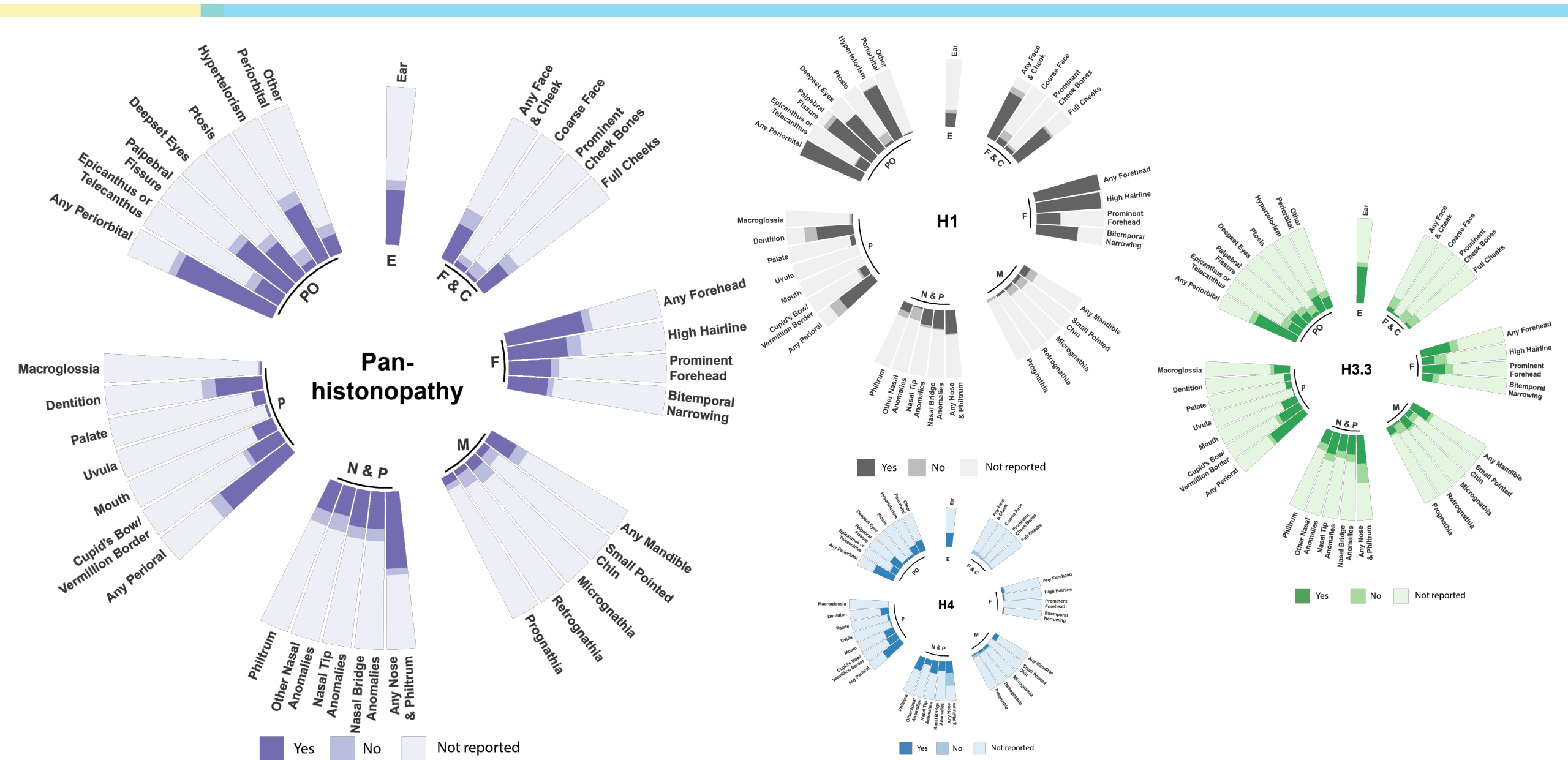
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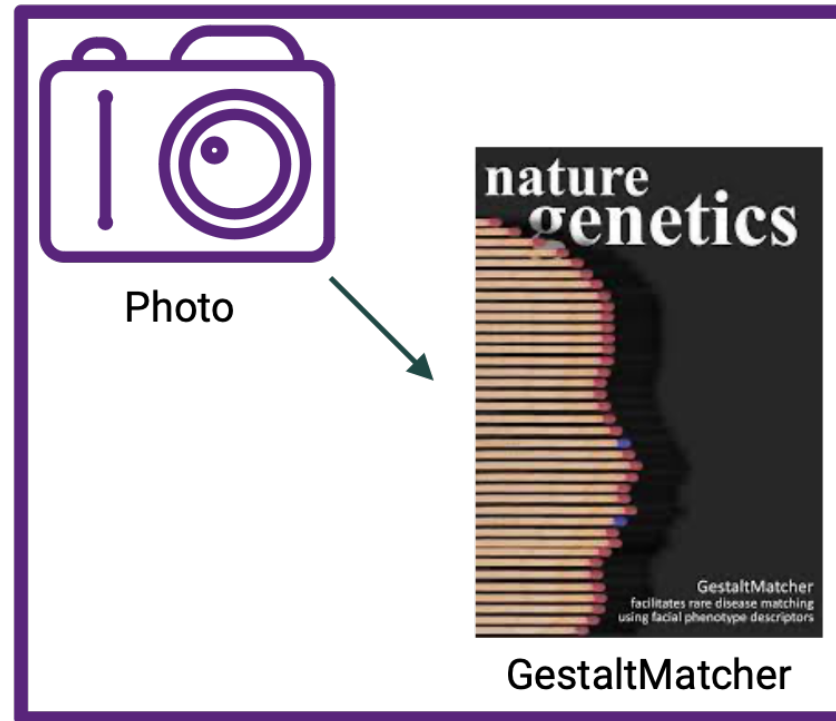


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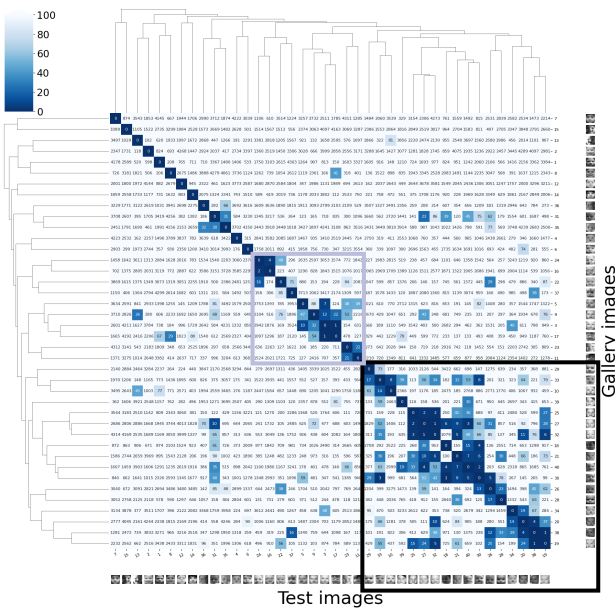


# Histone Syndromes – Leveraging AI for Histone-Based Syndromes

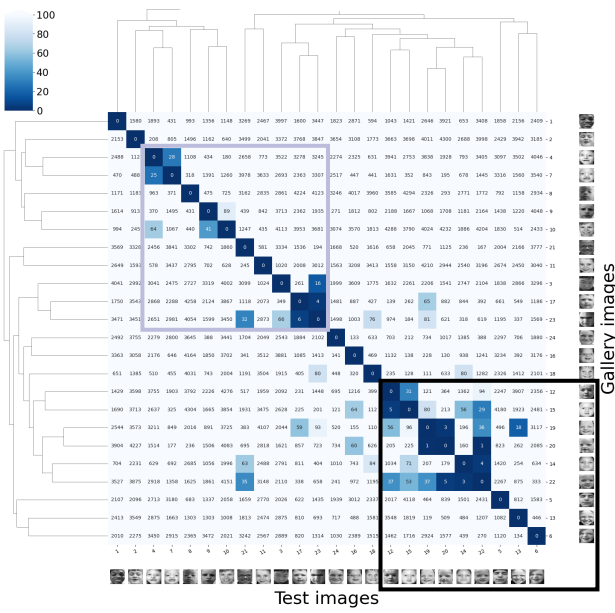


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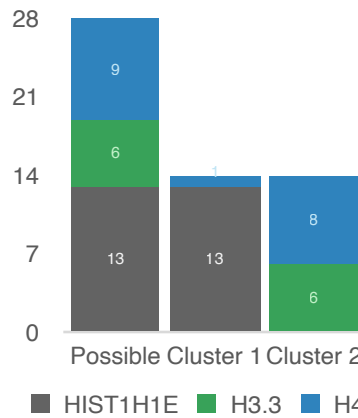
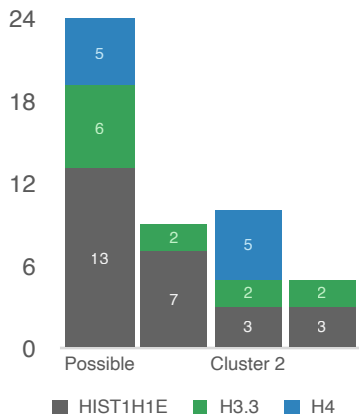
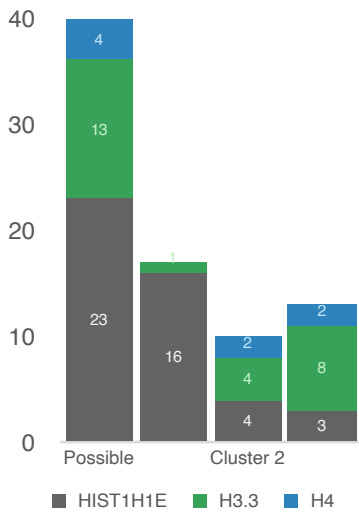
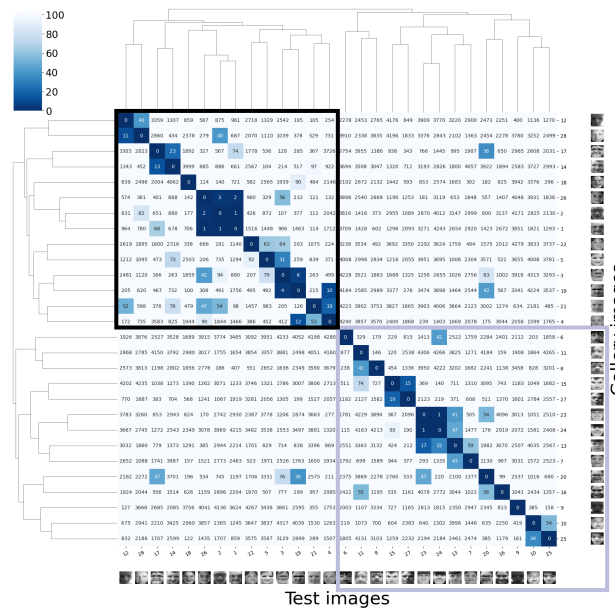
5 and under



6-12 year old



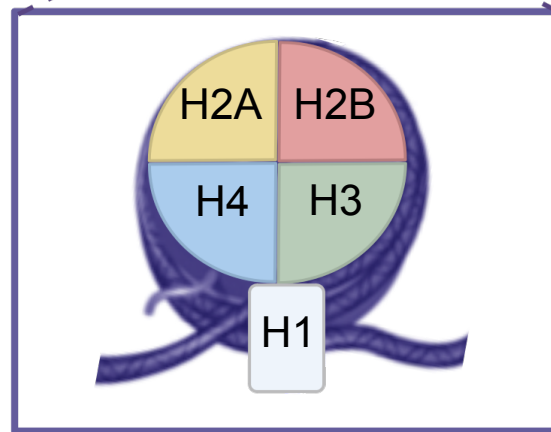
13 and over



# Moving Forward



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