Phenotype Chiari Malformation with Artificial Intelligence

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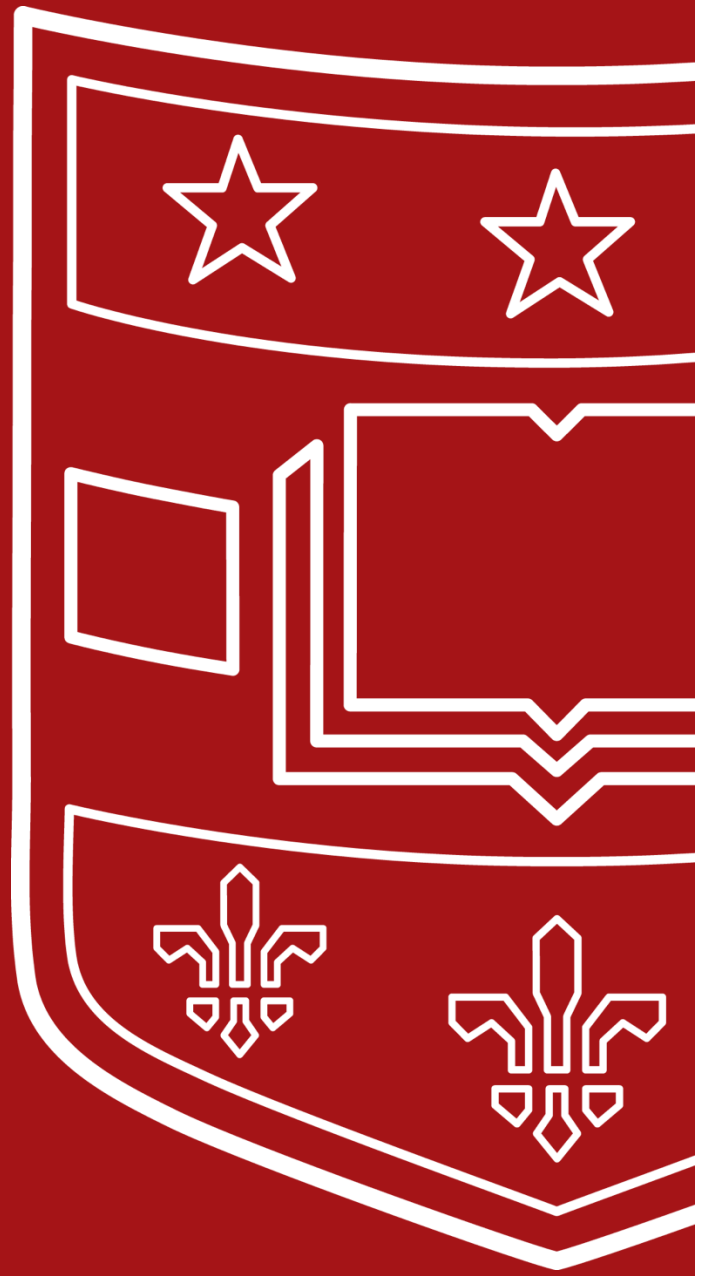


Washington University in St. Louis

Phenotype Chiari Malformation Type 1 (CM1)

- CM1 is a **heterogeneous** disease with significant variability in clinical presentation and natural history.
- There is growing recognition that CM1 likely represents a constellation of clinical phenotypes rather than a singular disease entity.
- Goal: Identify **distinct, clinically meaningful** phenotypes

Park Reeves Syringomyelia Research Consortium registry: 1340 patients from 42 medical centers since 2011

Orthopedic Info
e.g., Spinal Fusion

General Exam
e.g., Head Circumference

Symptoms
e.g., Neck Pain

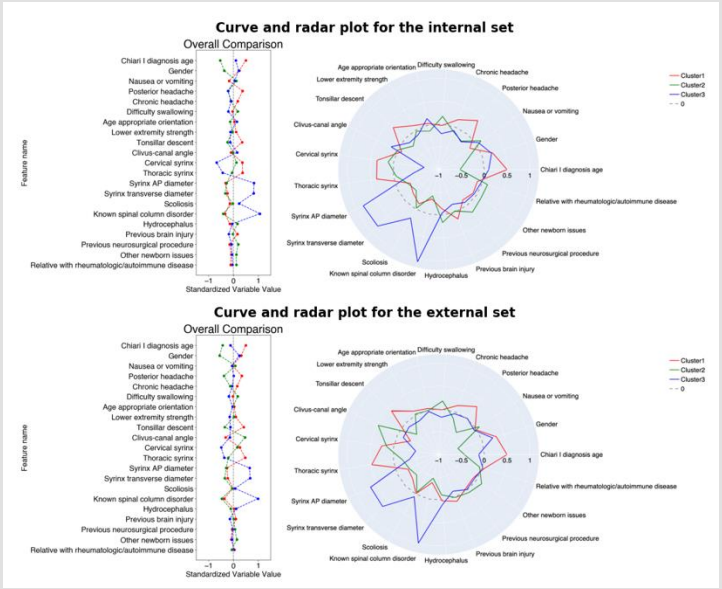
Input Features
(500+ variables)

Family History
e.g., Parent Syringomyelia

Childhood History
e.g., Chronic Headache

Radiographic Exam
e.g., Syrinx AP Diameter

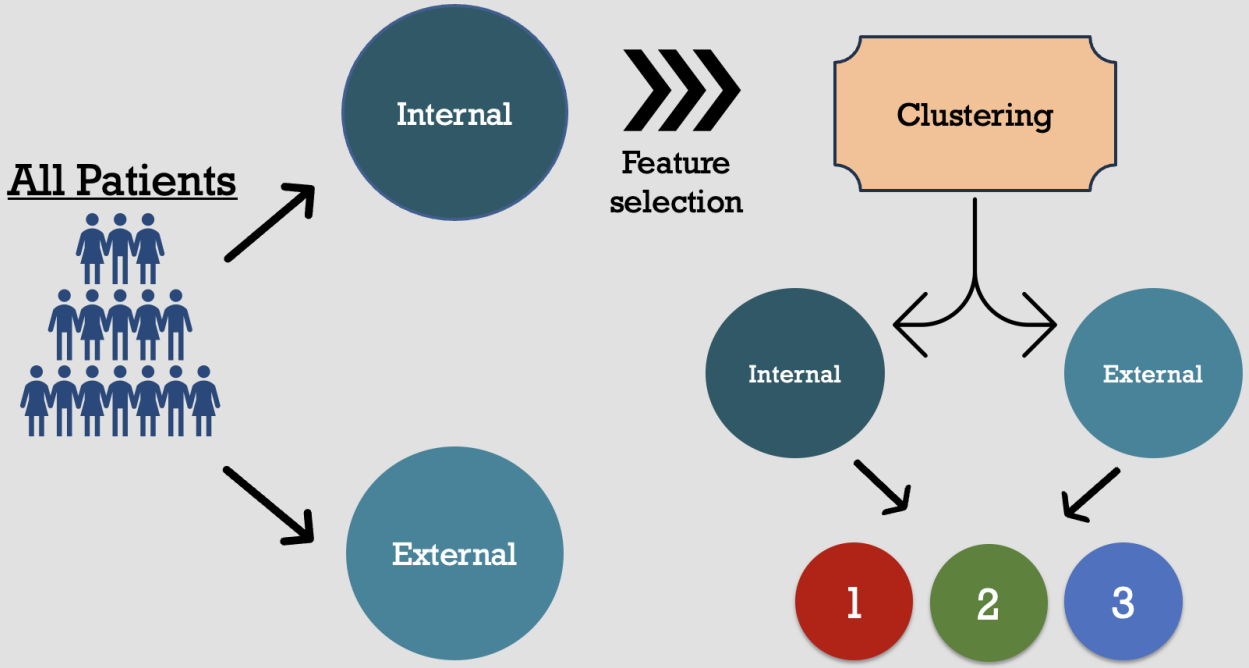
Neurological Exam
e.g., Facial Weakness



Work with Sean Gupta (Neurosurgery), Ziqi Xu (CSE), Jacob Greenberg, Dave Limbrick (Neurosurgery)

Modeling Approach

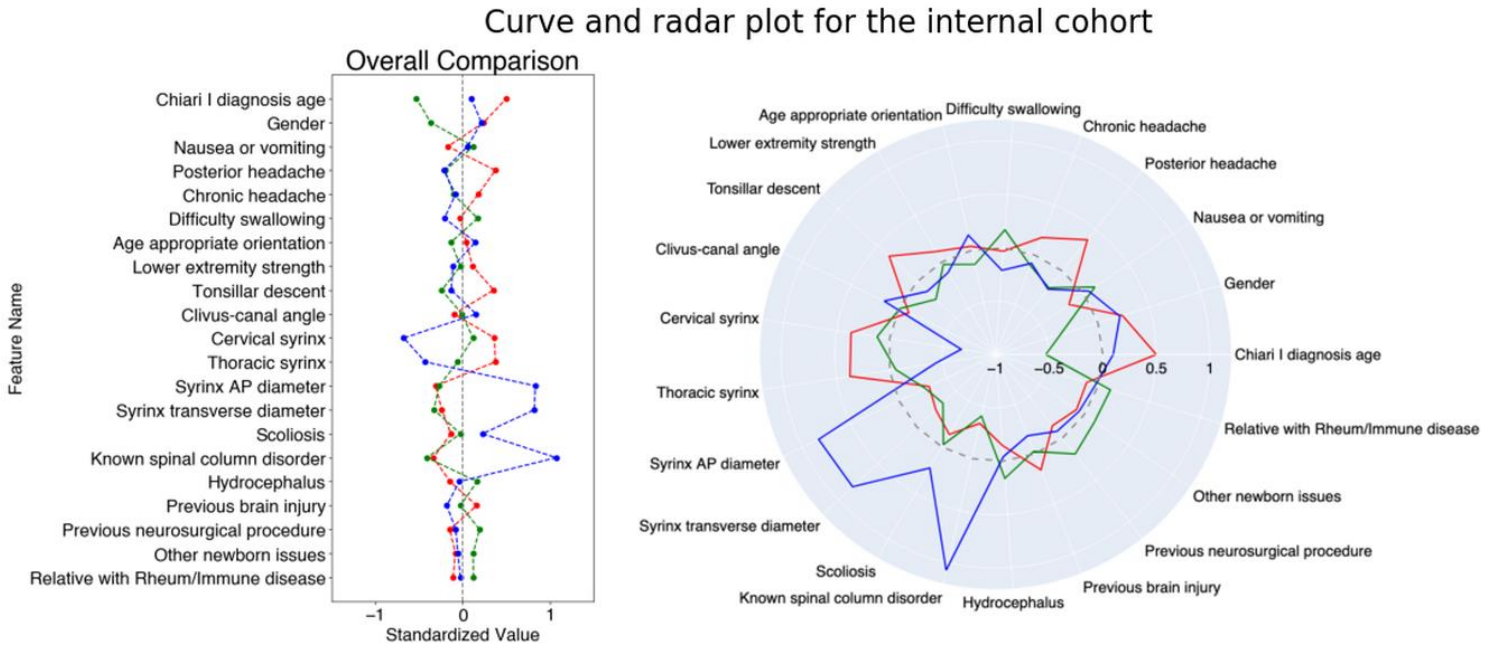
- Split 42 centers into internal and external cohorts
  - Test reproducibility
- Feature selection: identified 33 important predictors
  - 28 from data-driven methods
  - 5 from clinical surveys
- Unsupervised K-mode clustering

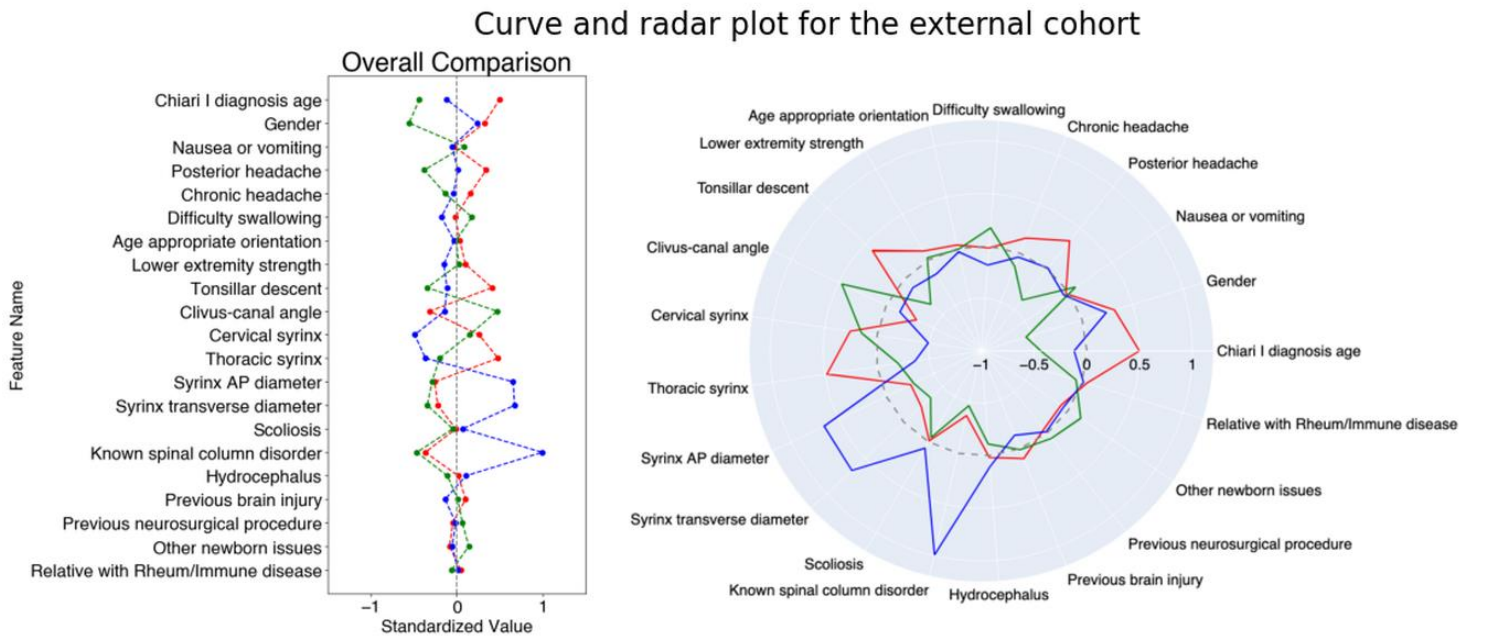


Clusters

- **Identified three distinct clusters with AI**
- **Reproducible in internal/external cohorts**

Gupta VP, Xu Z, Greenberg JK, Strahle JM, Haller G, Meehan T, Roberts A, Limbrick DD Jr, Lu C. Using artificial intelligence to identify three presenting phenotypes of Chiari type-1 malformation and syringomyelia. Neurosurgery. 2024 Nov 18.



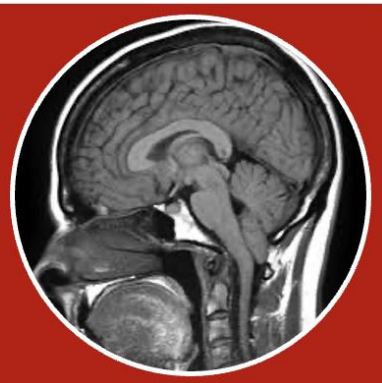


Clinical Implications

Cluster 1: Classic CM1+syrinx patients with primary headache complaints and few other symptoms or conditions.

Cluster 2: Patients with multiple neonatal medical issues but few classic CM1+syrinx symptoms—future symptom development under study.

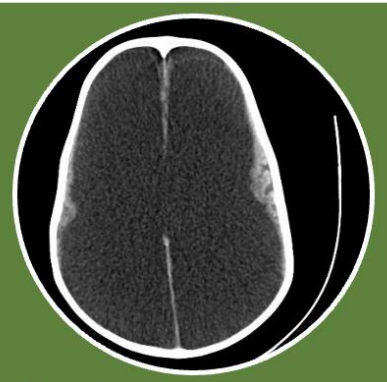
Cluster 3: CM1 patients with syrinx-related spinal effects; early diagnosis and intervention can help prevent major surgeries.



Cluster 1

- Highest diagnosis age
- More likely female
- Presents with HA
- Fewer bulbar symptoms
- Greatest tonsillar descent
- Small syrinxes
- Few other medical issues

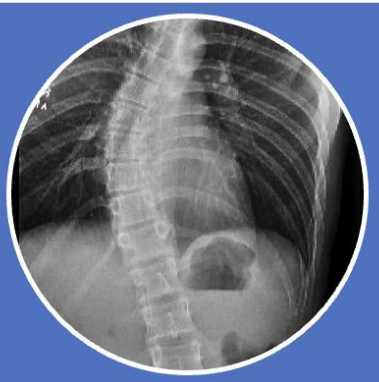
- n=277



Cluster 2

- Youngest diagnosis age
- Less tonsillar ectopia
- Small syrinxes
- Many newborn issues
- Fewer headaches
- Most hydrocephalus
- Most bulbar symptoms

- n=258



Cluster 3

- Less tonsillar ectopia
- Largest syrinxes
- Most spine disorders
- Most scoliosis
- Most motor deficits
- Fewer headaches
- Fewer bulbar symptoms

- n=187

- Typical radiologic definition is inadequate for heterogeneity of CM1
- Identified clusters potentially assist clinicians direct clinical management