Quantifying Severity in Craniosynostosis

A Machine Learning Approach With CranioRate™

Shireen Elhabian, PhD

Associate Professor of Computer Science

shireen@sci.utah.edu

University of Utah & University of Pittsburgh

NIDCR Grant R01-DE032366-01

Craniosynostosis

Premature suture fusion

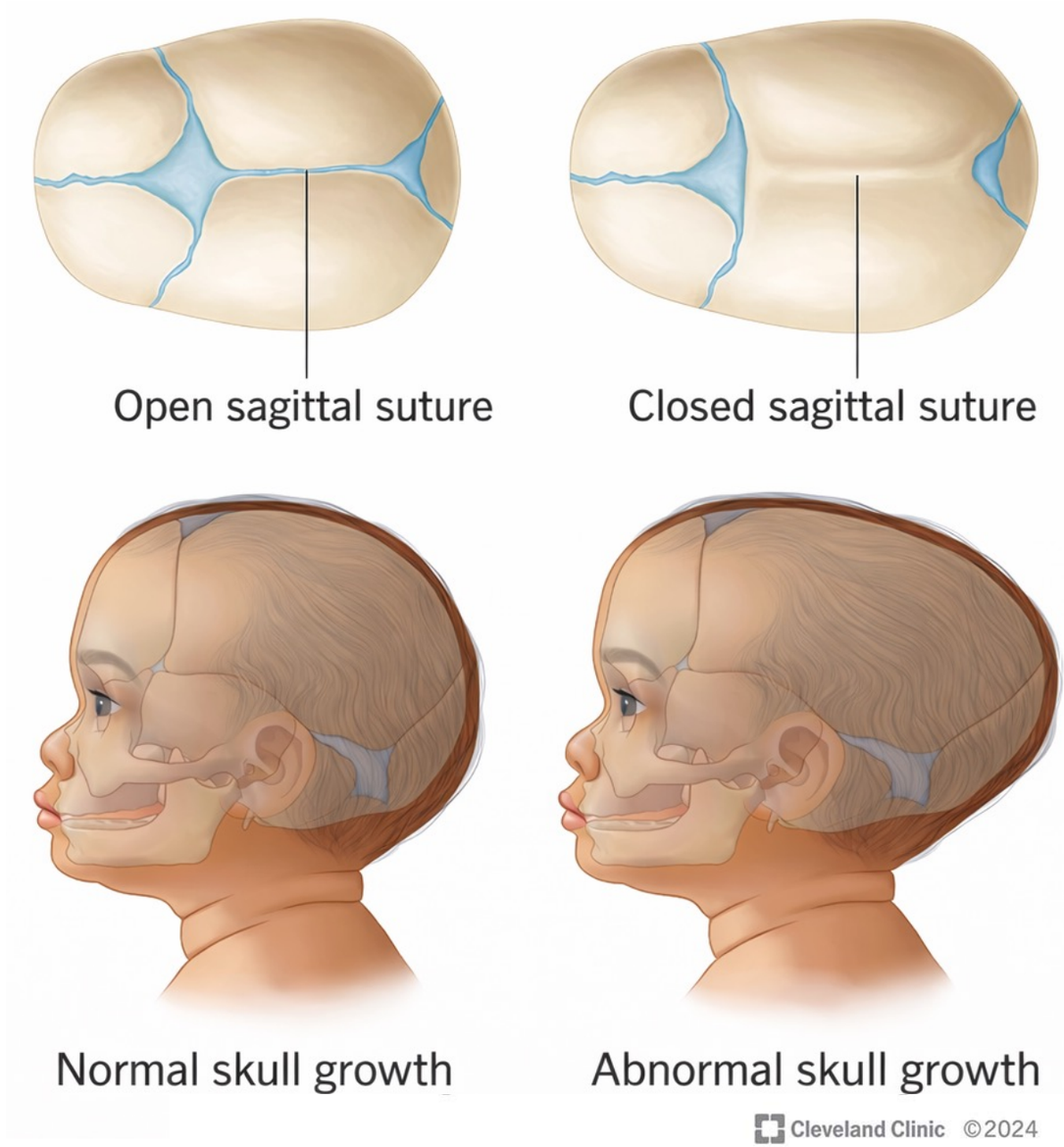
→ ~1 in 2,500 births

Abnormal head shape

Severity is subjective

Impacts surgical decision-making and outcomes

No standardized, quantitative definition of severity

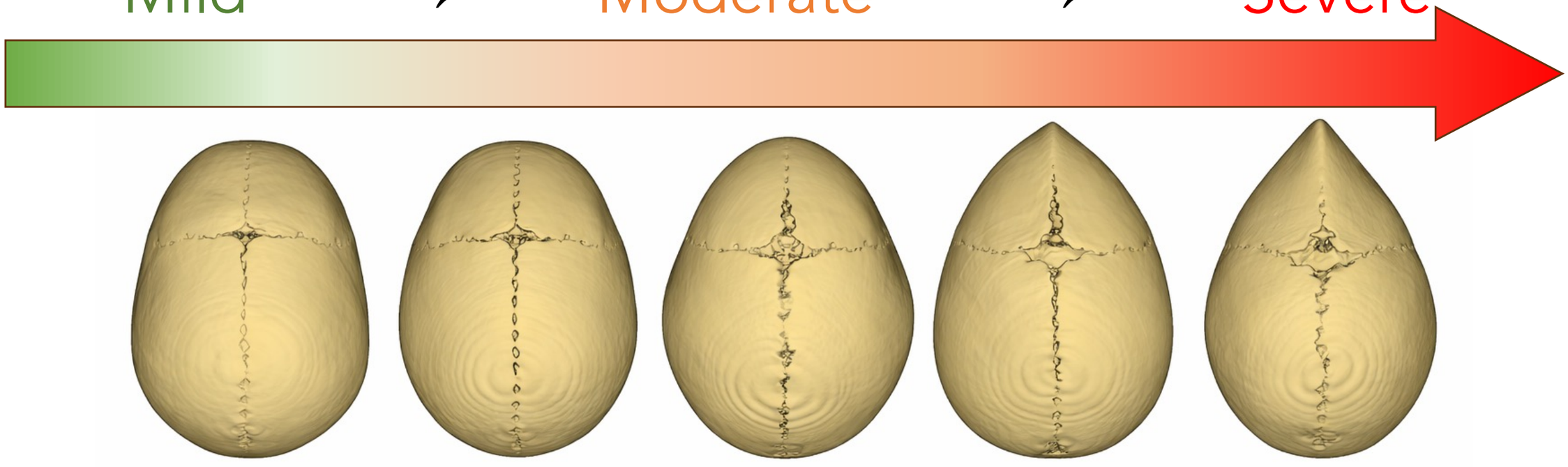


How do we measure severity objectively?

Craniosynostosis is a continuous spectrum

Not binary

Mild → Moderate → Severe

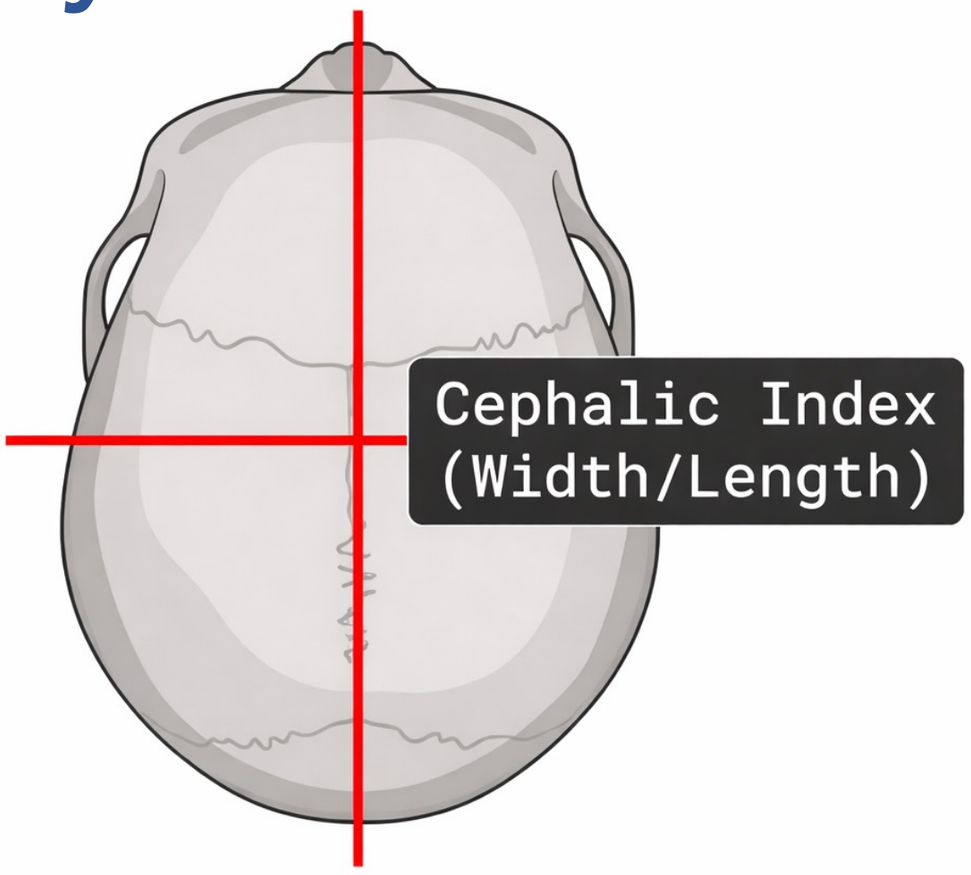


Where does a patient fall?

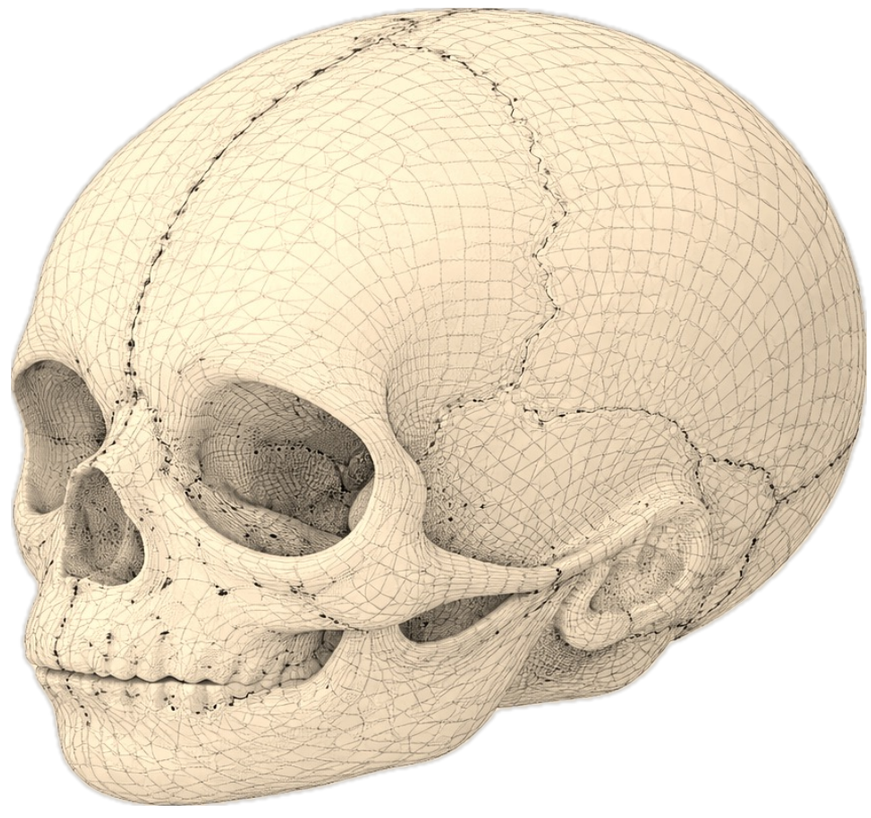
Current metrics cannot answer this reliably.

We need a continuous, quantitative representation of severity

Why current metrics fail to capture shape



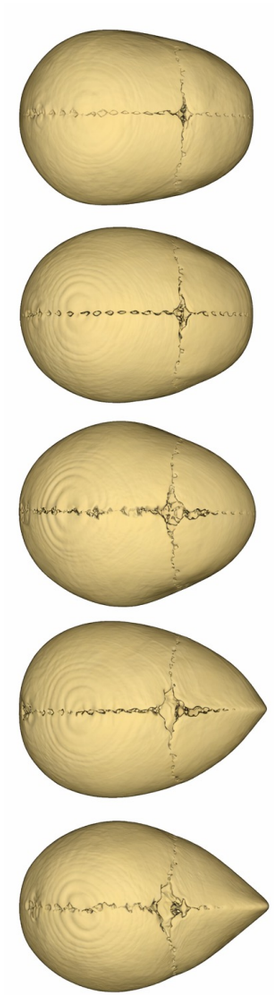
Reduces shape → single ratio
Ignores local variation
High inter-rater variability



Shape is continuous
High-dimensional
Requires full surface

Severity is not a line — it's a surface.

How clinicians assess severity



Prior cases (mental reference)

Education/Training

Clinical Experience (hundreds of cases)

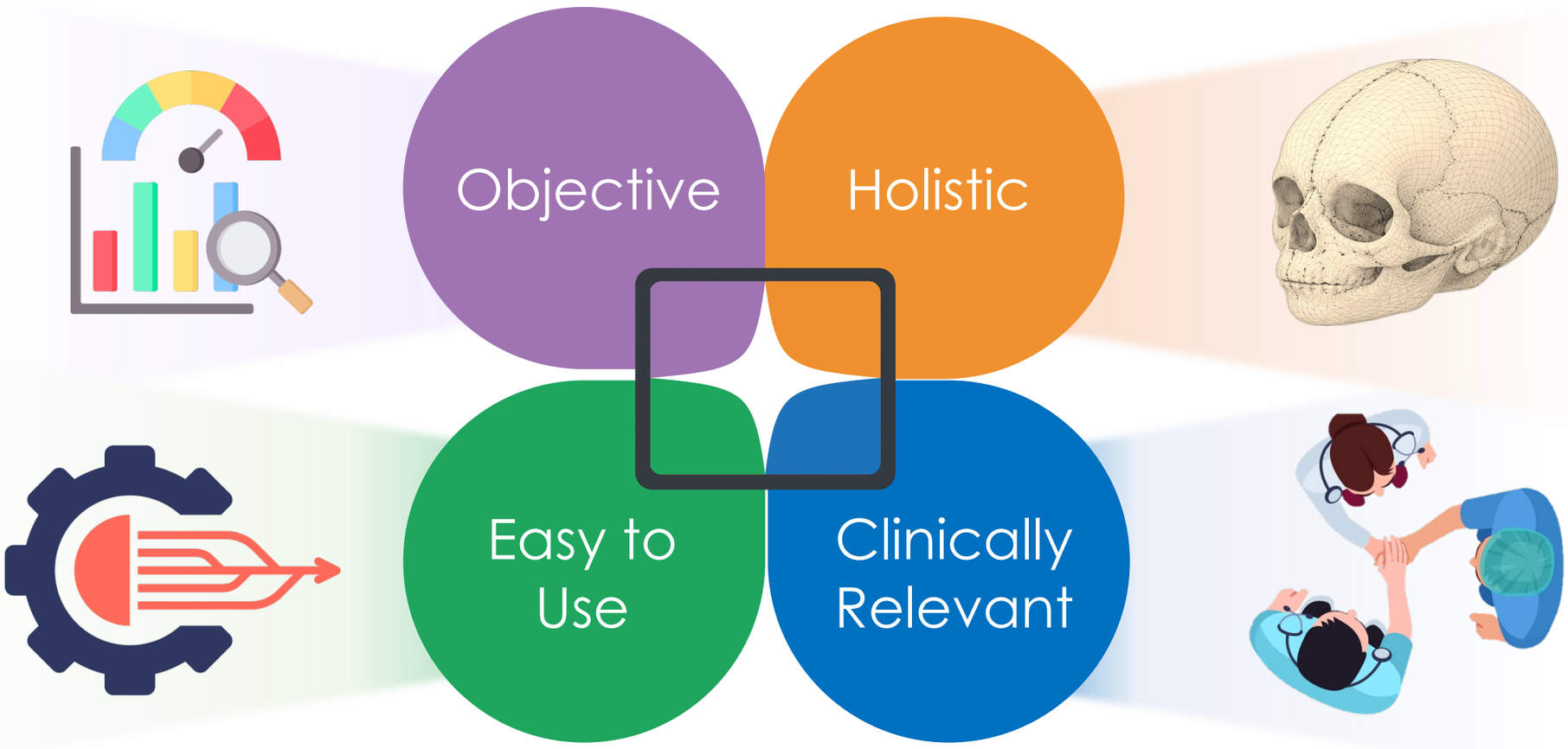
human pattern recognition over high-dimensional geometry

Diagnosis + Treatment decision

This process is powerful — but subjective and unstandardized.

Introducing CranioRate™

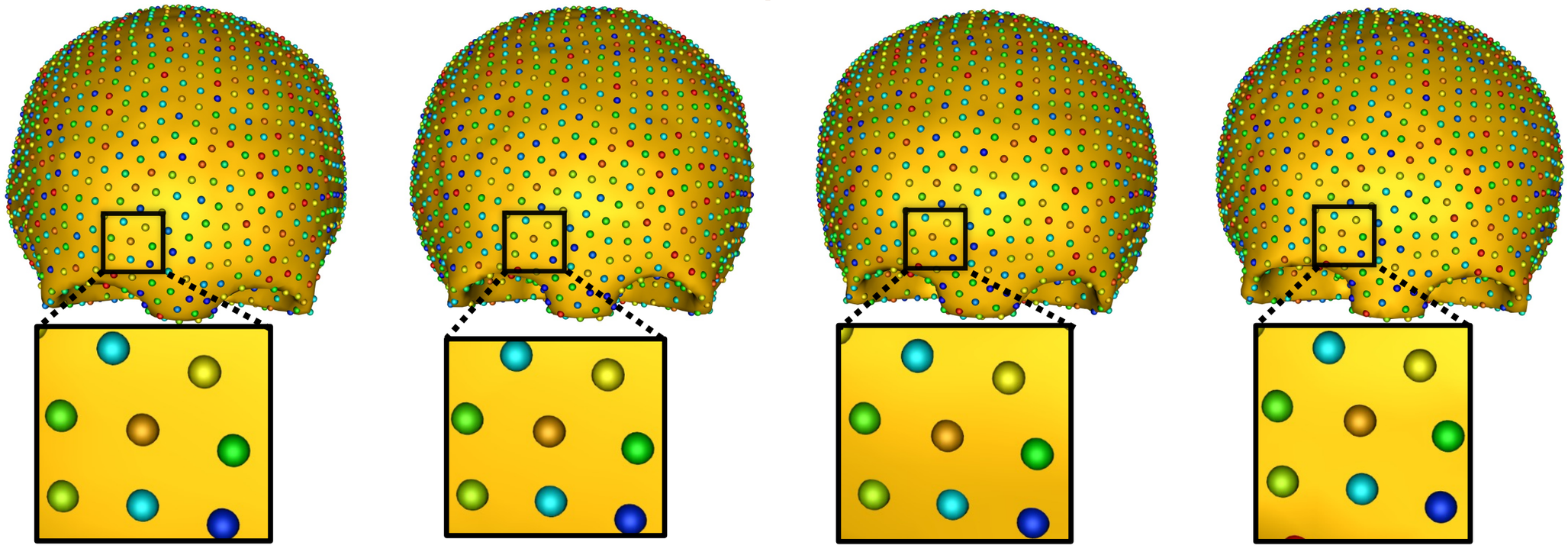
From subjective judgment to objective, reproducible measurement



A machine learning framework for quantitative cranial shape analysis

CranioRate™ – the secret sauce

Learning a standardized representation of anatomy across patients



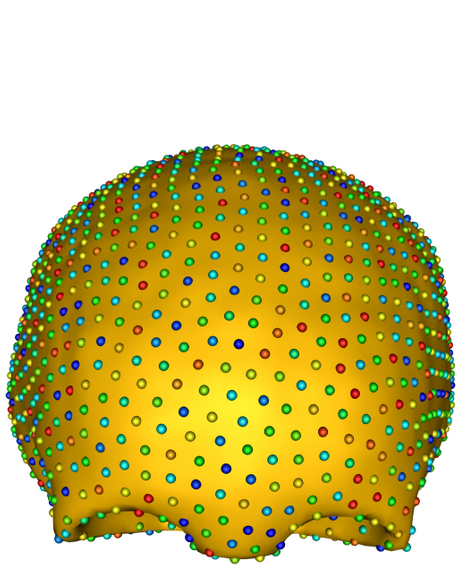
Same anatomical locations → same coordinates across patients

Enables population-level statistical learning

ShapeWorks
An Integrated Suite for Shape Representation and Analysis & more...

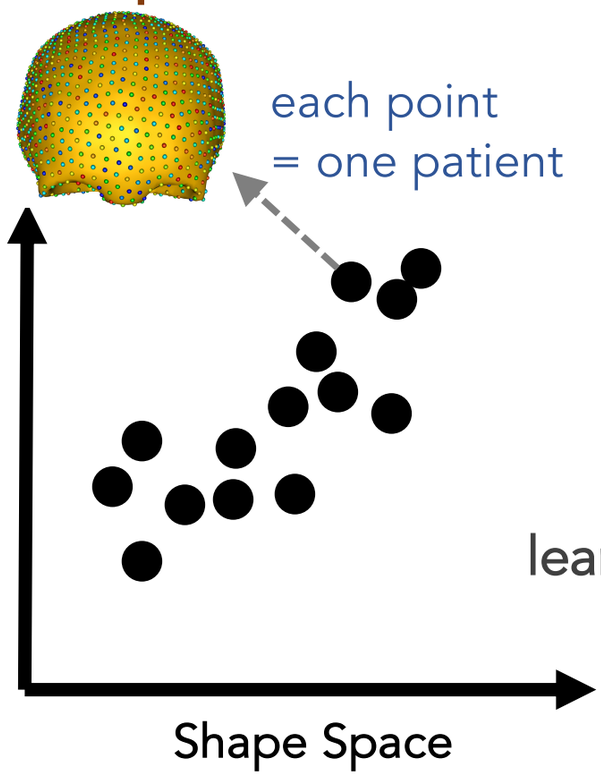
From anatomy to shape space

Each skull becomes a point in a quantitative shape space



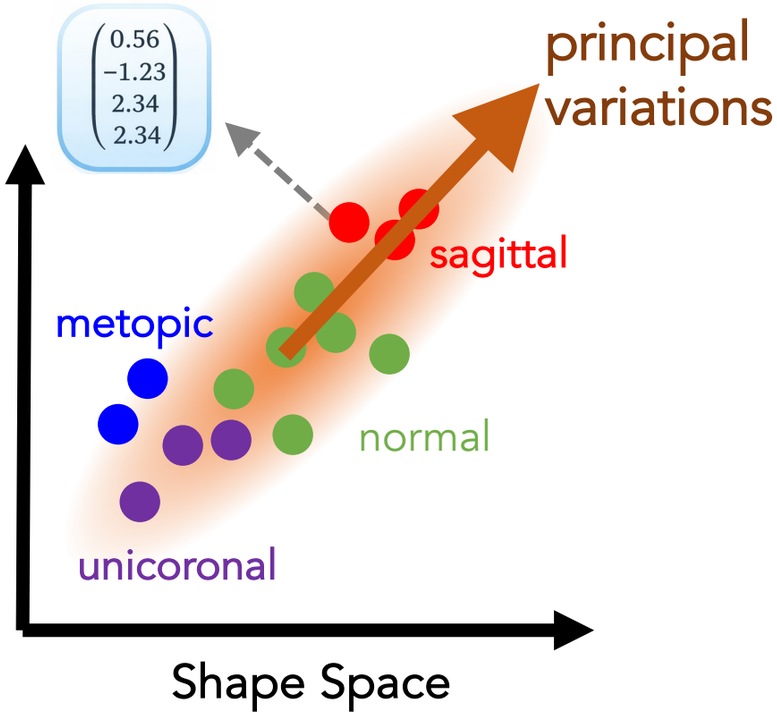
Corresponded Surface

encode shape as numbers



High-dimensional shape representation

learn dominant patterns



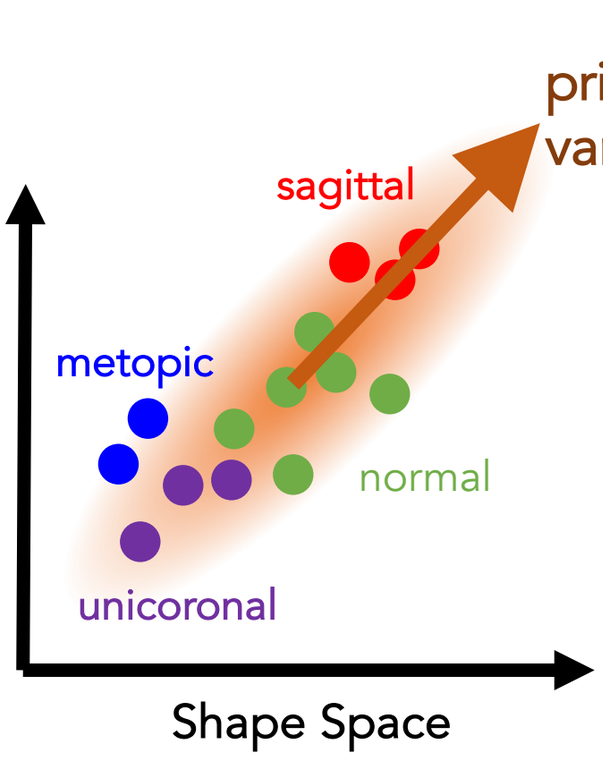
Low-dimensional morphological descriptor

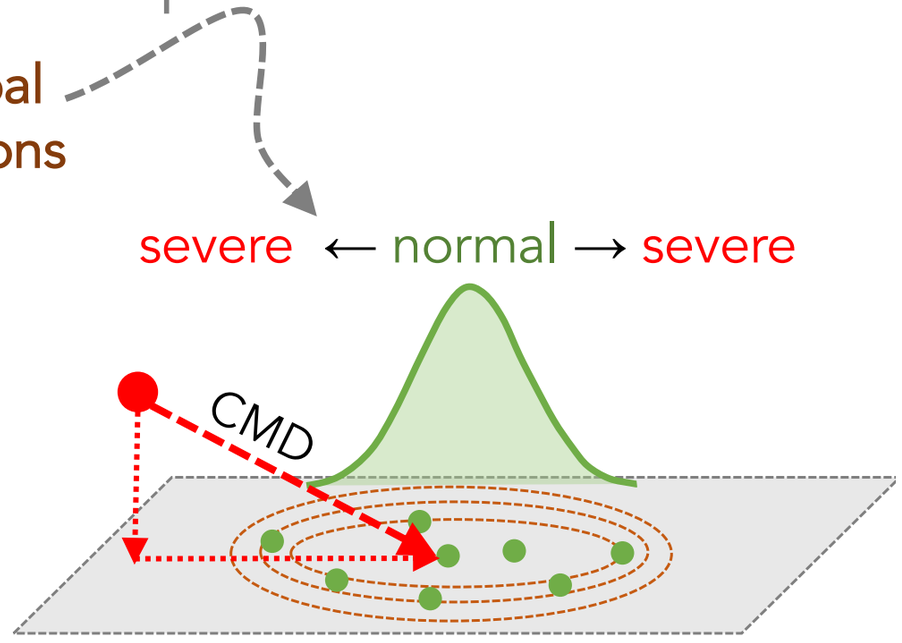
Normals cluster at the center → distance reflects severity

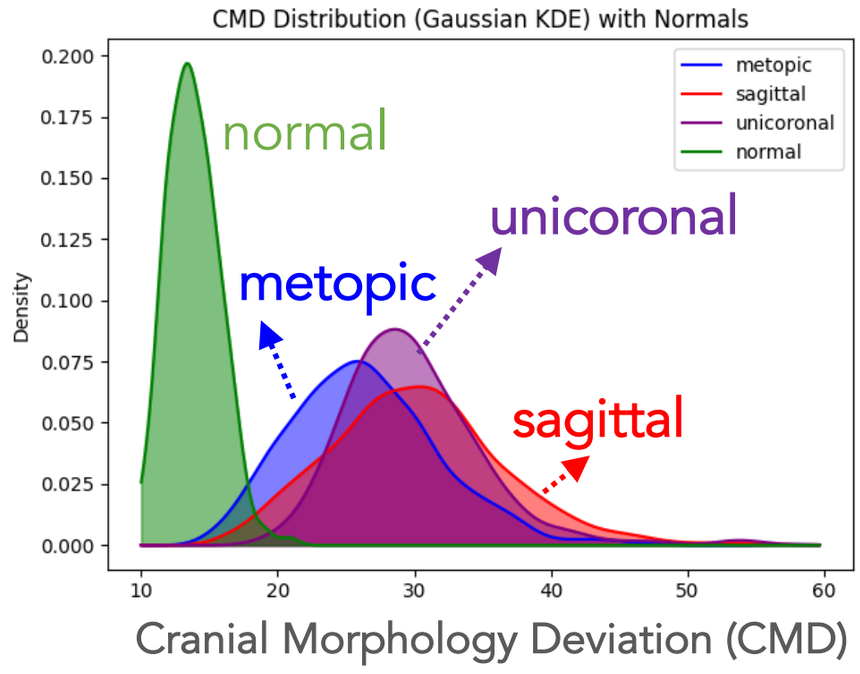
Quantifying deviation from normal shape

Unsupervised learning of normative anatomy

No labels required → robust to subjective bias







Cranial Morphology Deviation (CMD)
= deviation from normative model in the shape space

Validated in peer-reviewed studies (e.g., The Journal of Craniofacial Surgery, Plastic and Reconstructive Surgery)

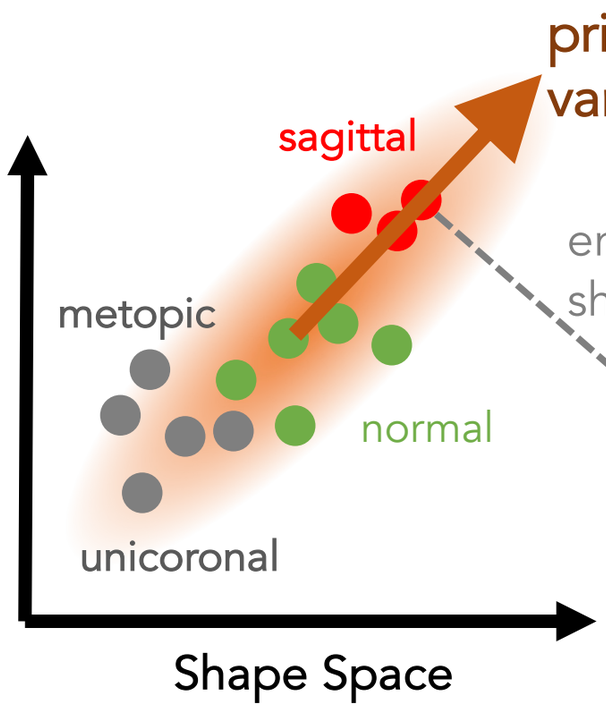
Expert-aligned severity scores

Trained on multiple expert ratings per patient

Maps shape to an objective, expert-aligned severity score

multiple expert ratings per patient

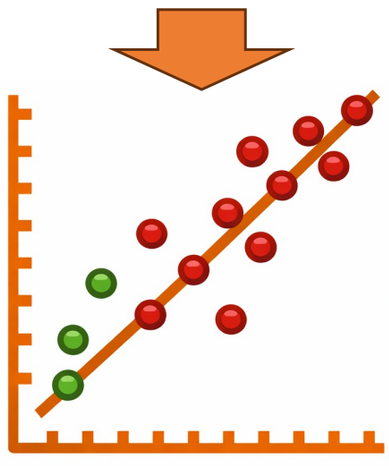
Learning from noisy, subjective expert labels



encode shape

$$\begin{pmatrix} 0.56 \\ -1.23 \\ 2.34 \\ 2.34 \end{pmatrix}$$

learn mapping



Item Response Theory (IRT) Model

Probabilistic model of rater variability

predict severity

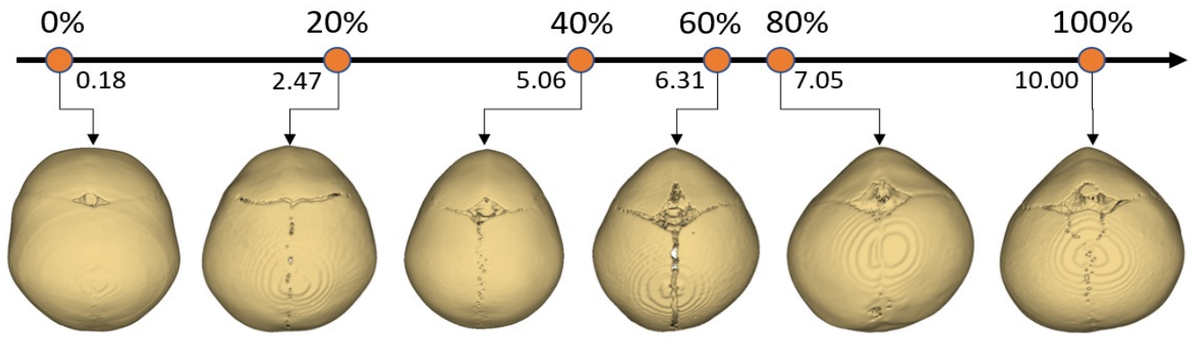
Sagittal Severity Score

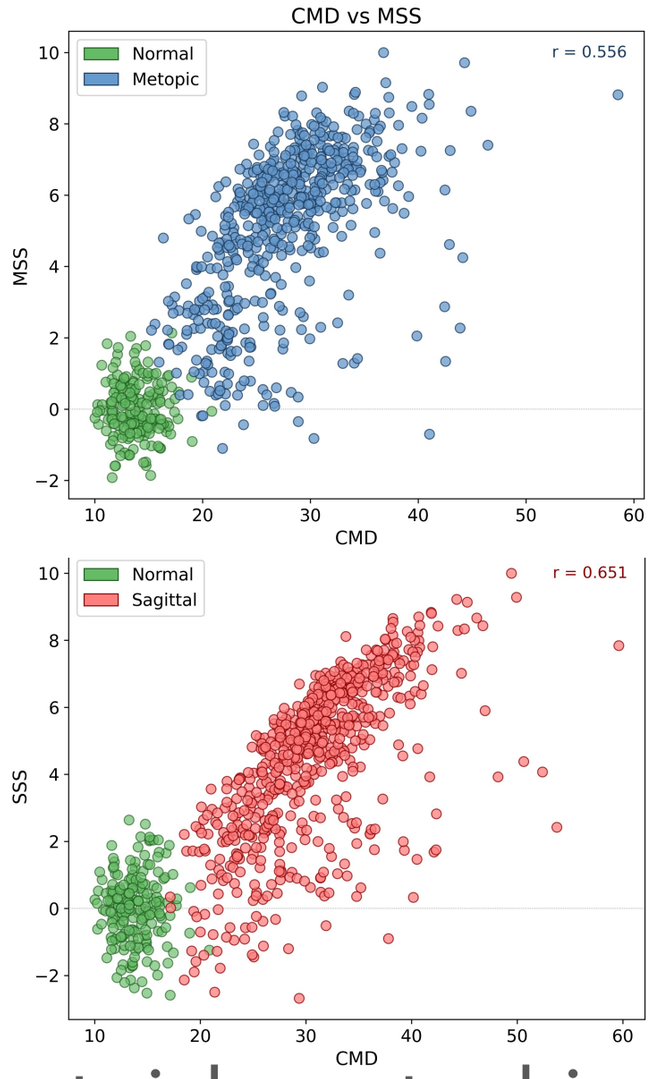
Red (active): sagittal

Gray: other phenotypes (context only)

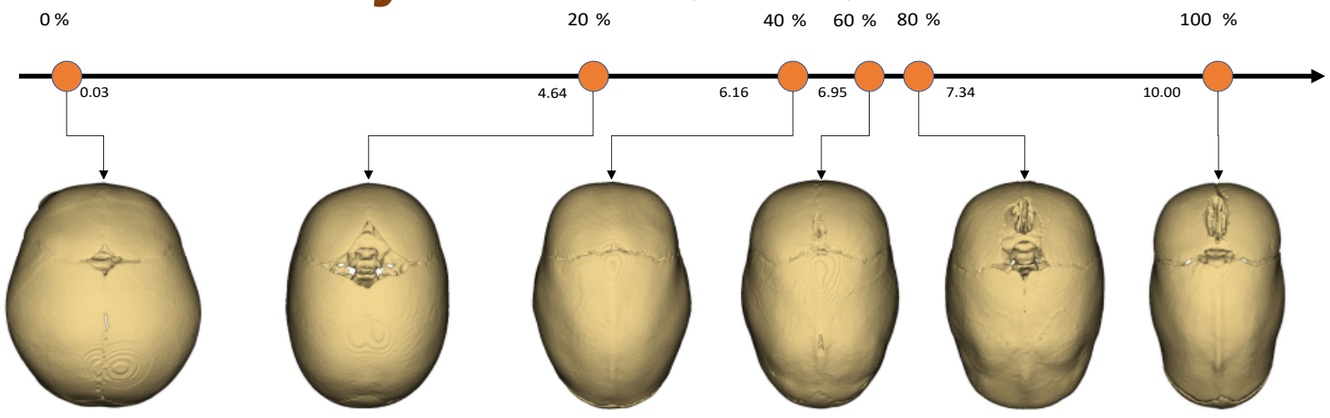
Evaluation: Agreement with expert-defined severity

Metopic Severity Score (MSS)





Sagittal Severity Score (SSS)



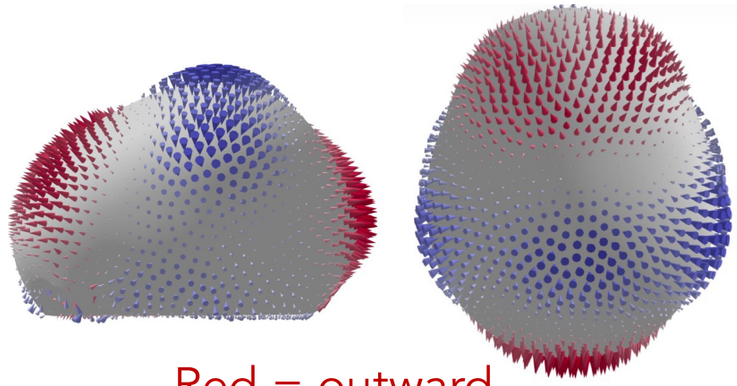
Higher CMD → higher MSS / SSS

Strong correlation but not perfect → reflects inherent subjectivity

Validated in peer-reviewed studies (e.g., The Journal of Craniofacial Surgery, Plastic and Reconstructive Surgery)

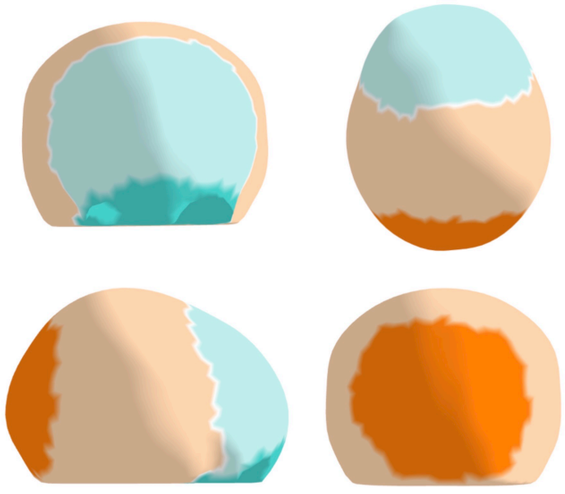
Interpretability: linking ML scores to anatomical regions

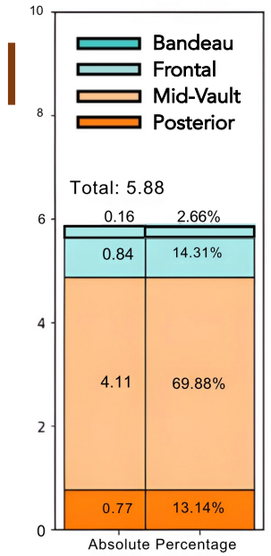
Severity reflects spatial deformation patterns

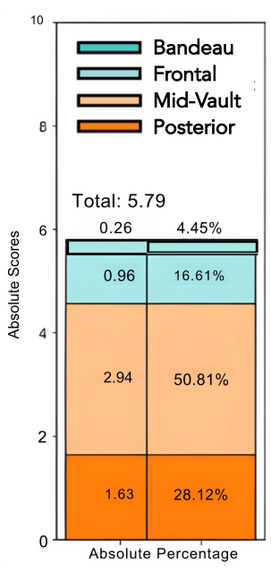


Red = outward

Blue = inward



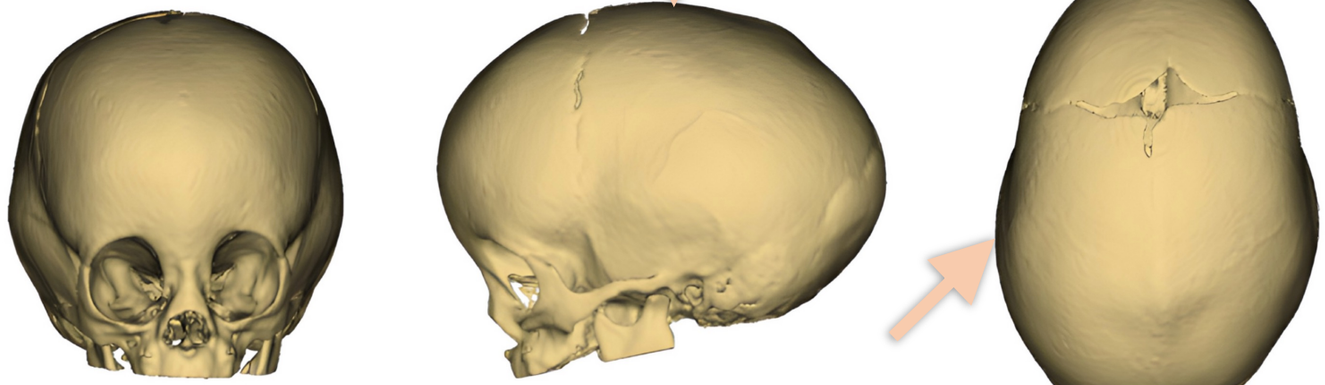




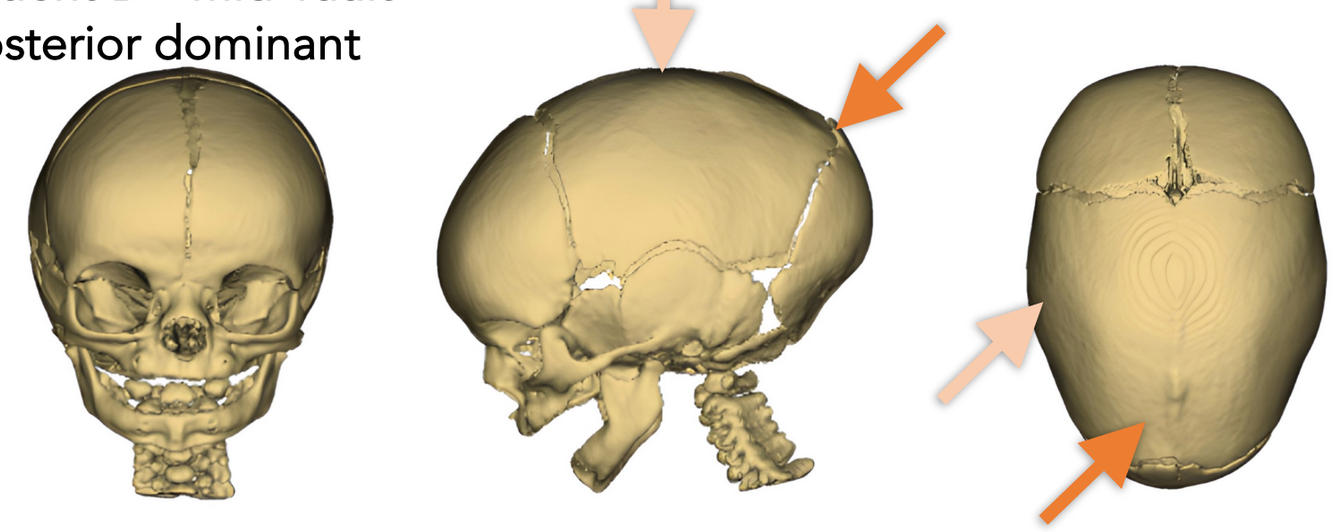
Bandeau
Frontal
Mid-Vault
Posterior

Regional contribution to severity

Patient A – mid-vault dominant



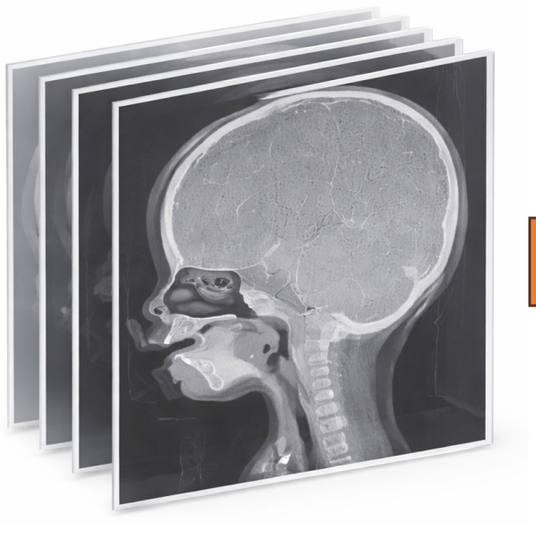
Patient B – mid-vault + posterior dominant



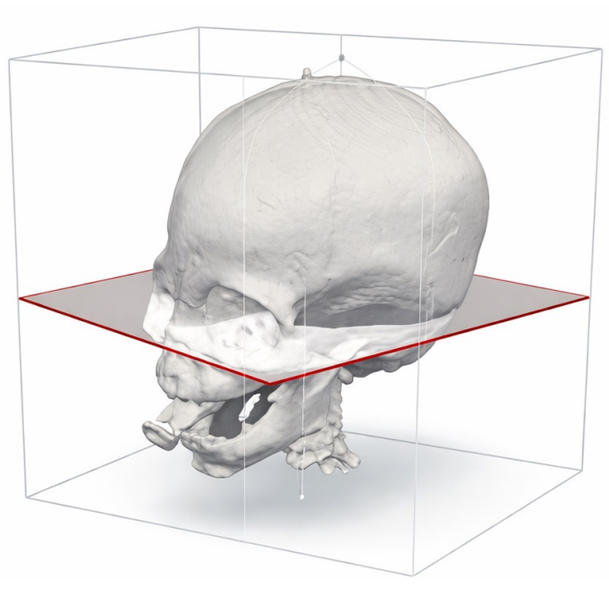
Patients with similar severity can have different anatomical drivers

From imaging to scores – fully automated

End-to-end ML pipeline from raw imaging to quantitative phenotype— no manual steps required



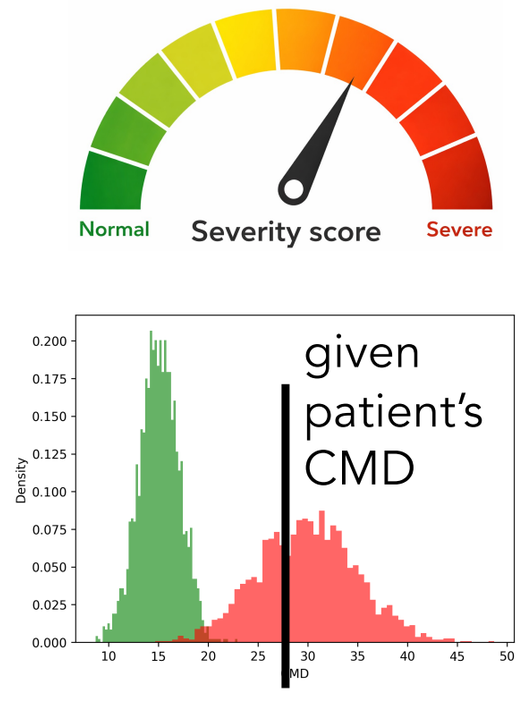
CT Scan



3D Cranium Extraction

Shape Representation

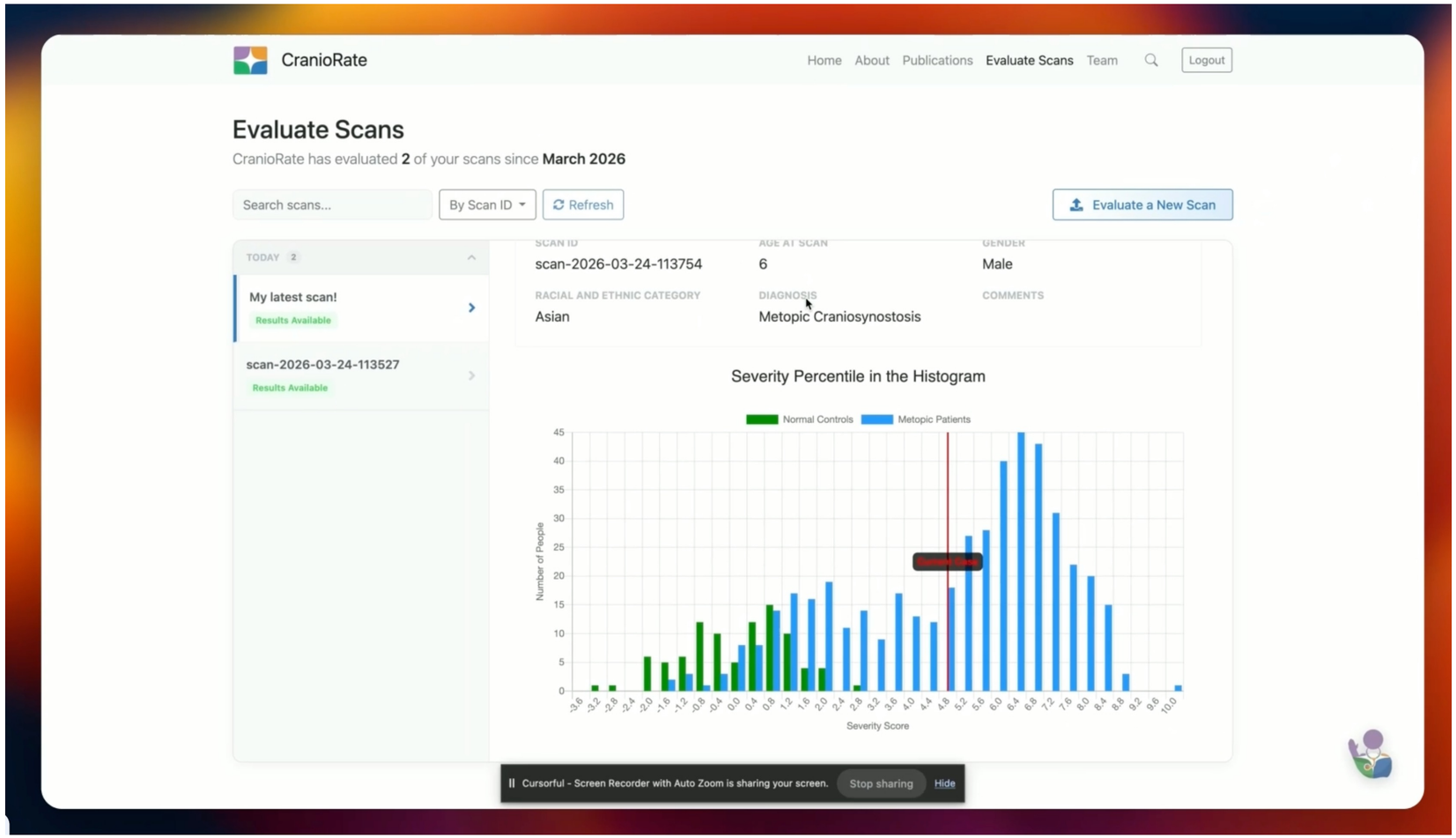
(patient encoded in shape space)



Quantitative Scores

(CMD & Severity)

CranioRate™ at the Point of Care

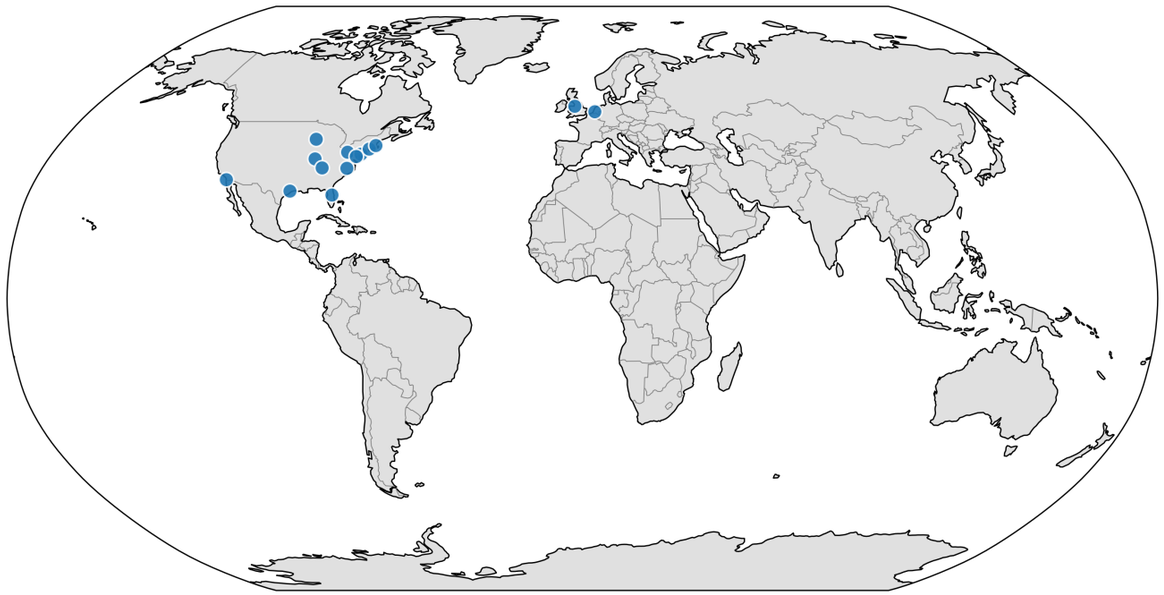


Upload a CT → Receive quantitative severity scores

Translating ML models into clinical decision support tools

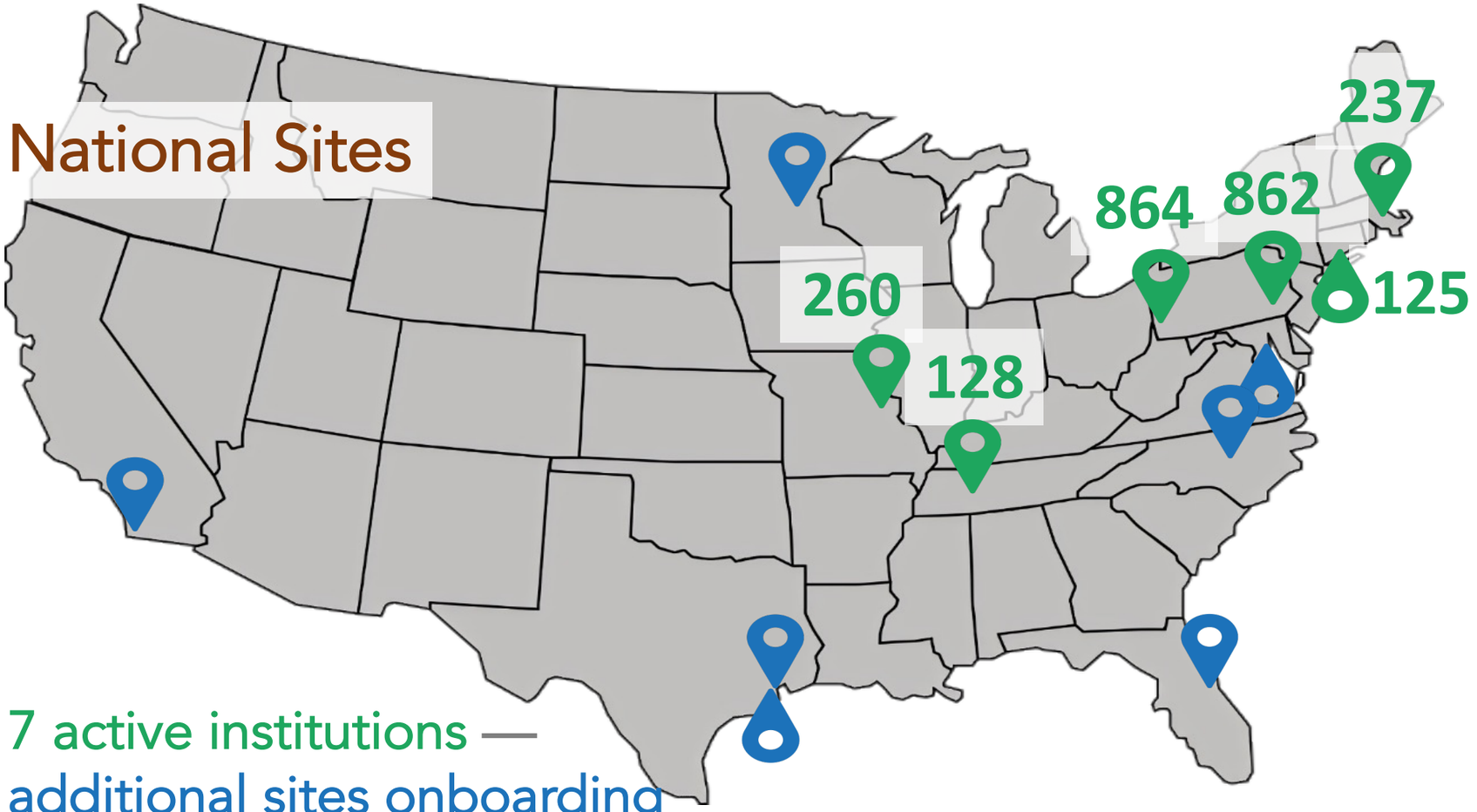
Point of care in action

Providing quantitative assessment across multiple institutions



No patient data is stored or used for model training

CranioRate Consortium & Bulk Processing at Scale

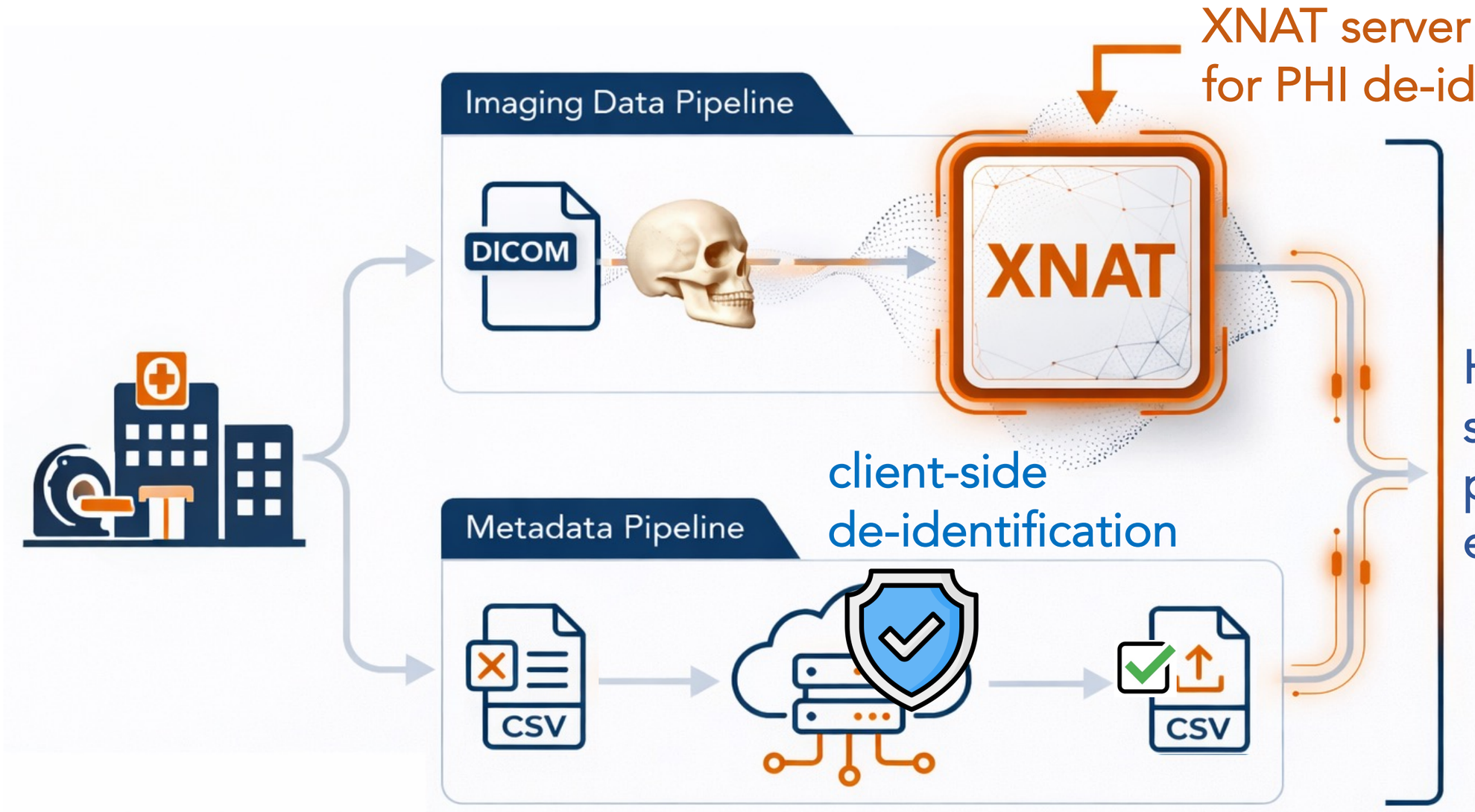


Total: 2901 Scans Uploaded & Processed

Jan 2025 – Present

Enables efficient batch processing and identification of edge cases to improve robustness

Secure data flow for consortium uploads

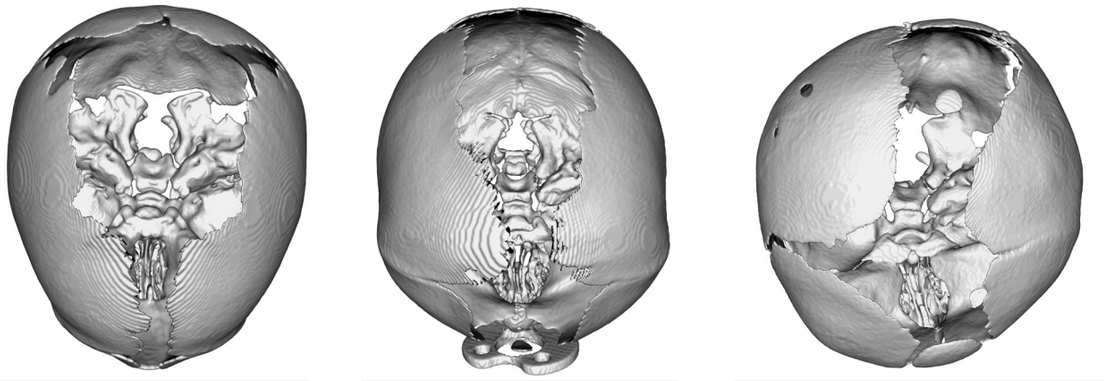


Standardized, secure pipelines are required for reproducible ML

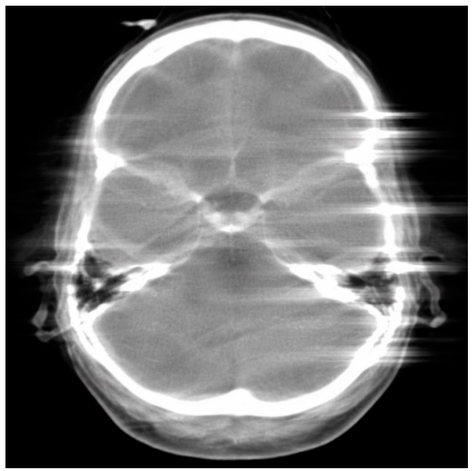
Secure bulk upload infrastructure for consortium sites

Real-world challenges in clinical AI

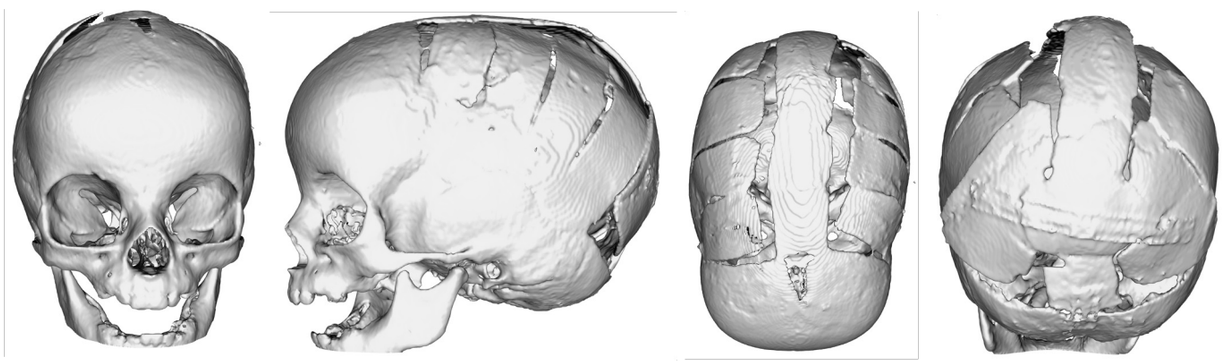
~13% pipeline failure rate in real-world clinical scans



Younger patients (thin/soft bone) → low contrast → ambiguous boundaries → incorrect severity



Motion artifacts → blurred edges → thresholding failure → incorrect severity

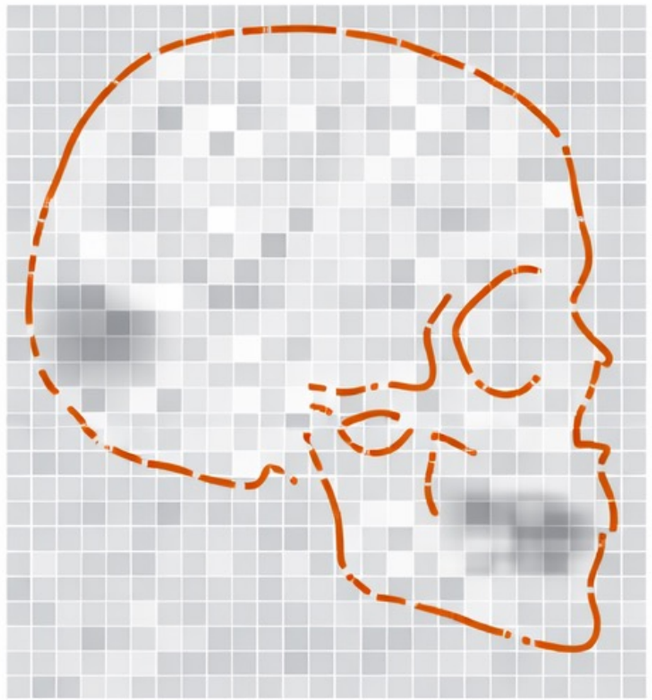


Post-operative scan → artifacts + false/missing structures → incorrect severity

Noisy CT scans → unstable thresholding → surface artifacts → incorrect severity

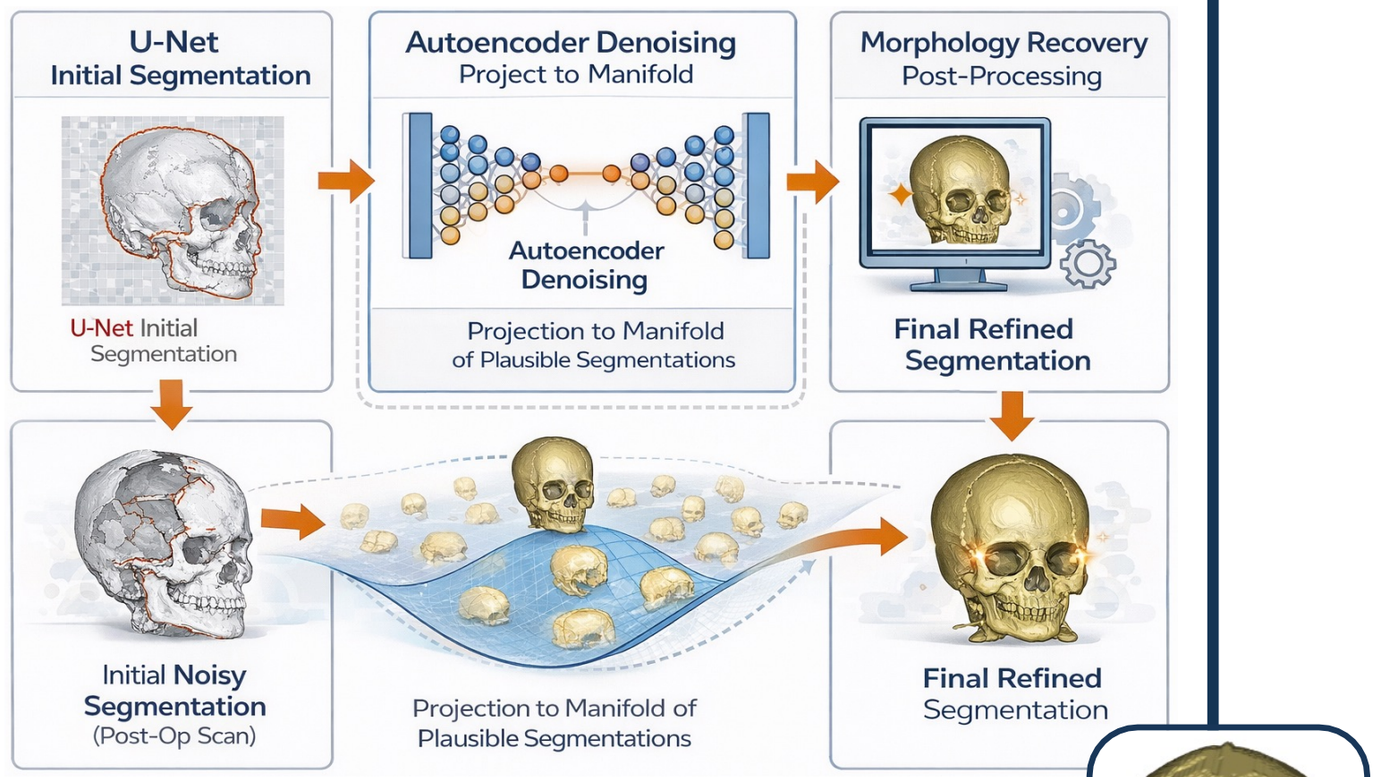
CranioRate 2.0: Anatomy-aware segmentation

V1.0 Thresholding



Fails on challenging scans (soft bones, motion, post-op, noise)

V2.0 Deep Learning

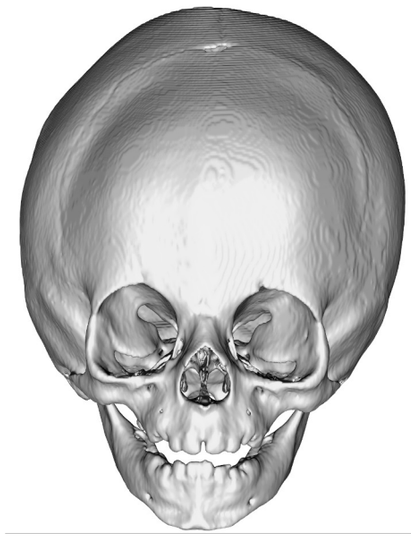
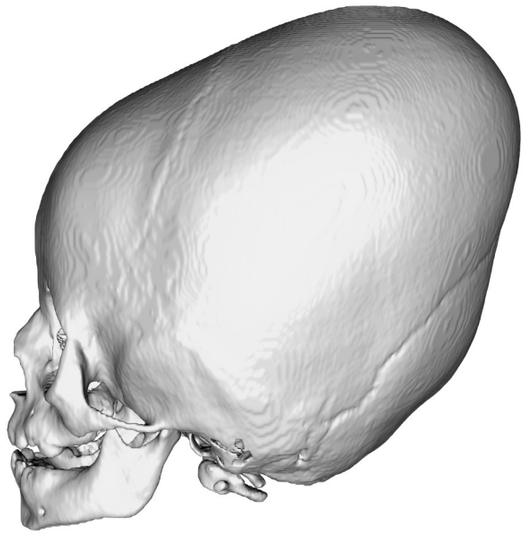
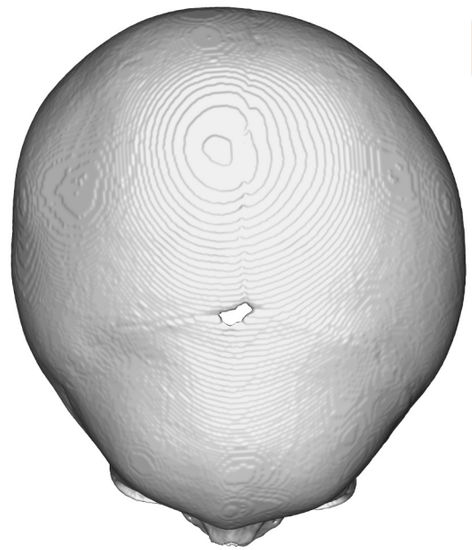


Deep learning segmentation → robust across diverse scan conditions

Shape-aware refinement → corrects artifacts and enforces anatomical plausibility

Generalizes across sites

CranioRate 2.0: Improved severity quantification

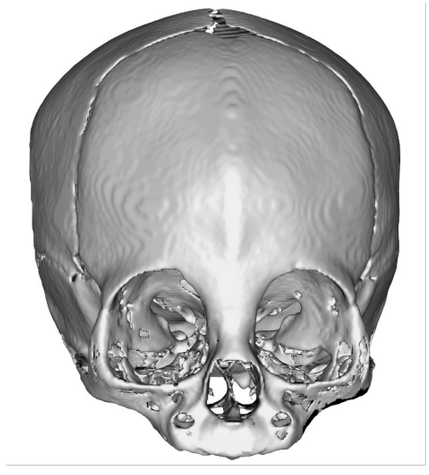
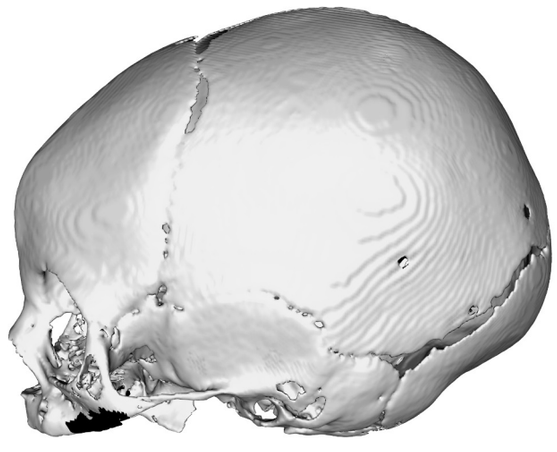
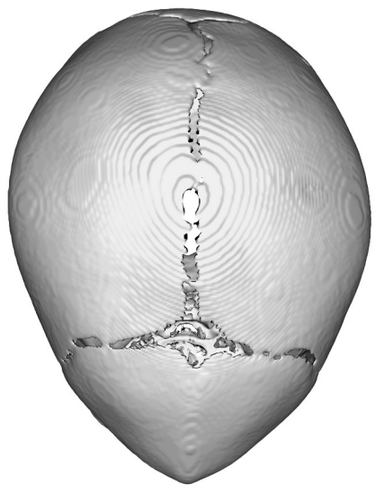
Metopic craniosynostosis

MSS v1.0 - 5.017

↓

MSS v2.0 - 9.716 ↑

moderate → severe

MSS v1.0 - 3.568

↓

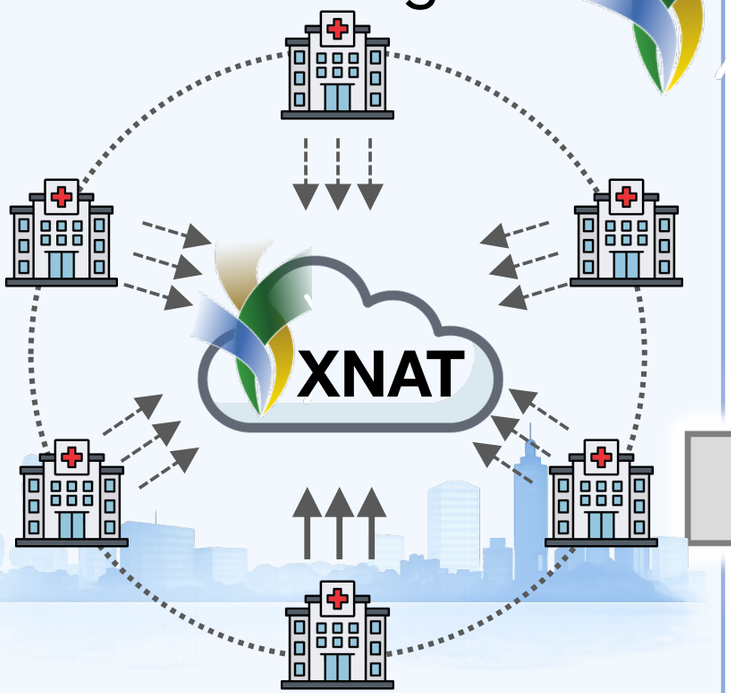
MSS v2.0 - 6.904 ↑

mild → near severe

Correcting systematic underestimation in challenging clinical scans

Scaling impact: Structured data sharing

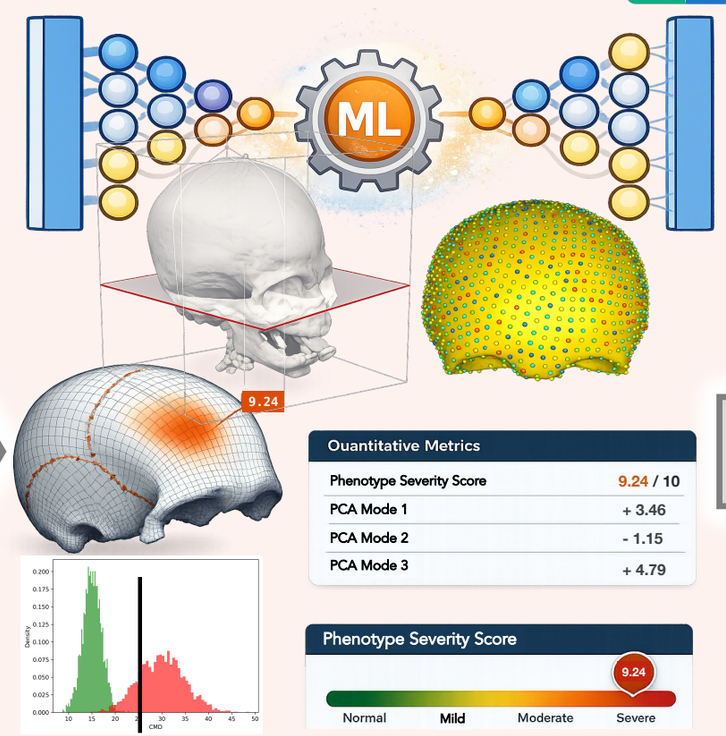
Multi-site Data Ingestion



WashU Medicine · Children's Hospital of Philadelphia · UPMC · Children's Hospital Boston · Erasmus MC University Medical Center Rotterdam · VANDERBILT UNIVERSITY

Bulk uploads from collaborating clinical sites

CranioRate Processing



Quantitative Metrics	
Phenotype Severity Score	9.24 / 10
PCA Mode 1	+ 3.46
PCA Mode 2	- 1.15
PCA Mode 3	+ 4.79

Phenotype Severity Score: 9.24 — Normal · Mild · Moderate · Severe

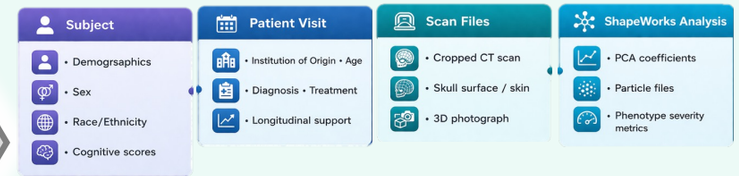
Transforms raw scans into standardized, quantitative cranium anatomy for analysis and sharing

FaceBase

A Resource For Craniofacial Researchers

NIDCR-funded craniofacial repository

Clinicians · Researchers · ML / AI · Consortiums



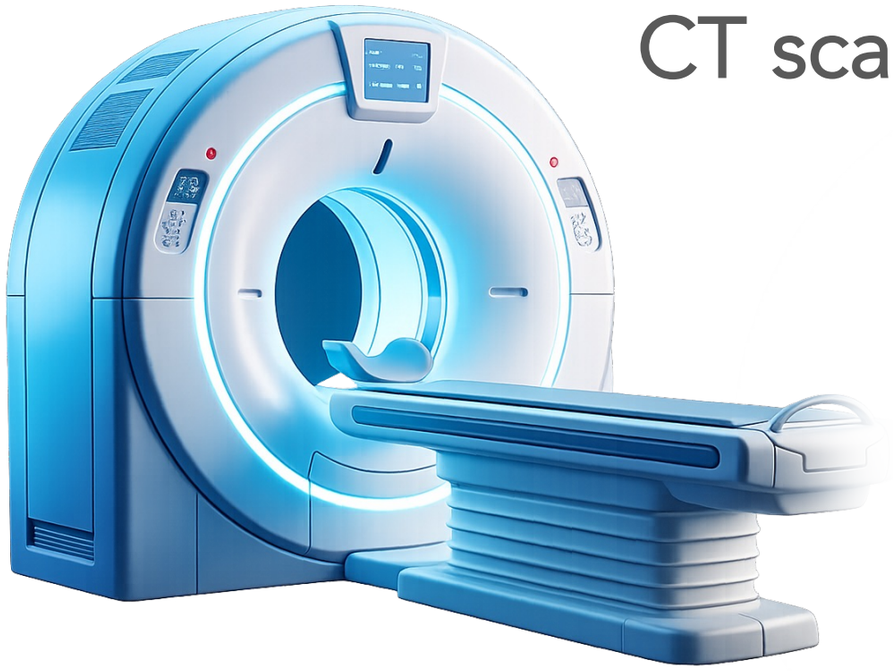
FAIR Principles · Open Access · NIDCR Backed

- API-based data submission
- Longitudinal versioning
- Secure, controlled research access

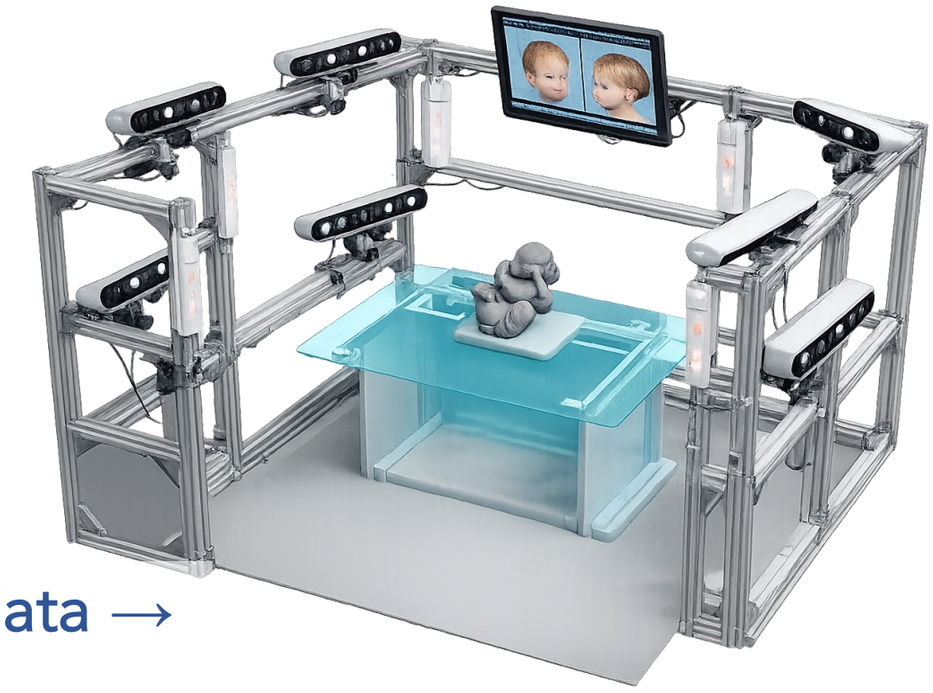
Standardized representations + FAIR data enable scalable clinical AI

From CT to scalable 3D imaging

CT scanning | 3D stereophotogrammetry



THE SHIFT



From high-quality, sparse data → scalable, noisier data

- ✓ High precision
- ⚠ Radiation exposure limits longitudinal use

- ✓ Radiation-free
- ✓ Fast, repeatable acquisition
- ✓ Scalable across clinics

From radiation imaging to safe, repeatable, population-scale measurement

… but

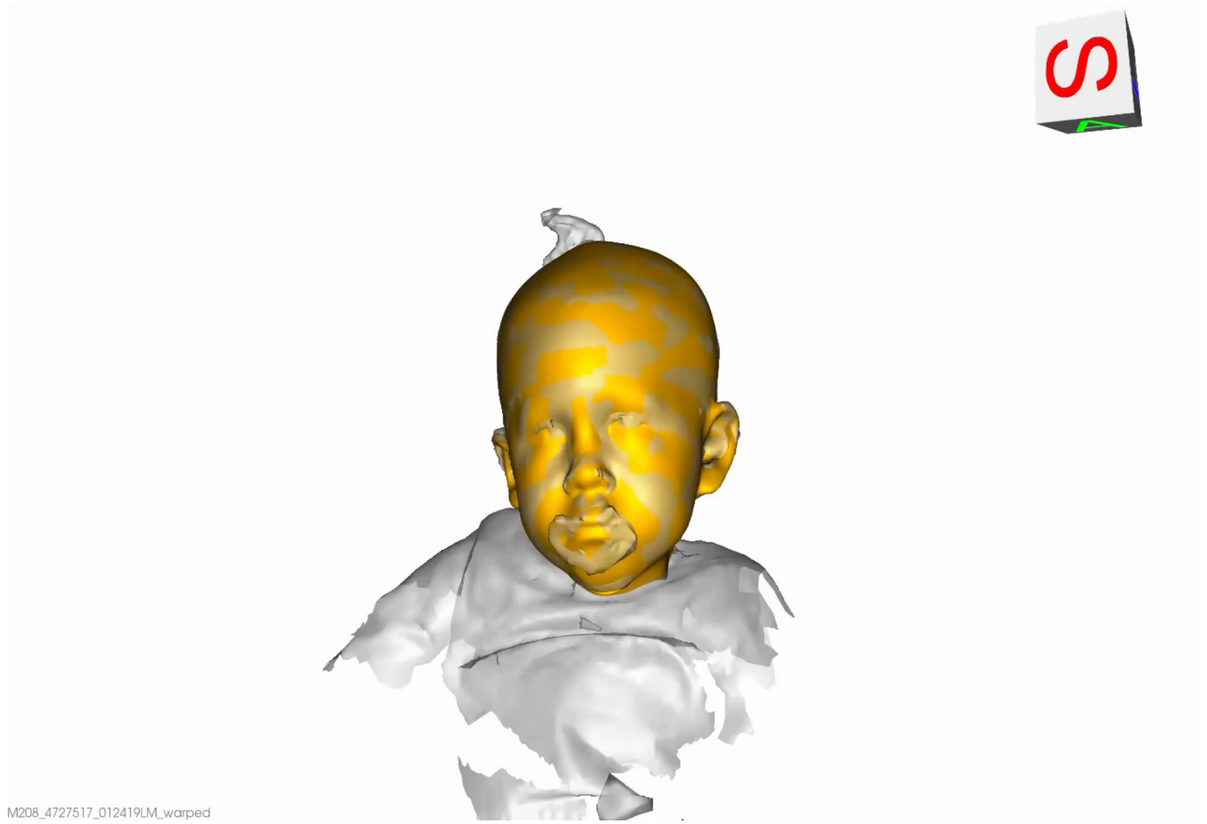
✗ Noisy & incomplete surface

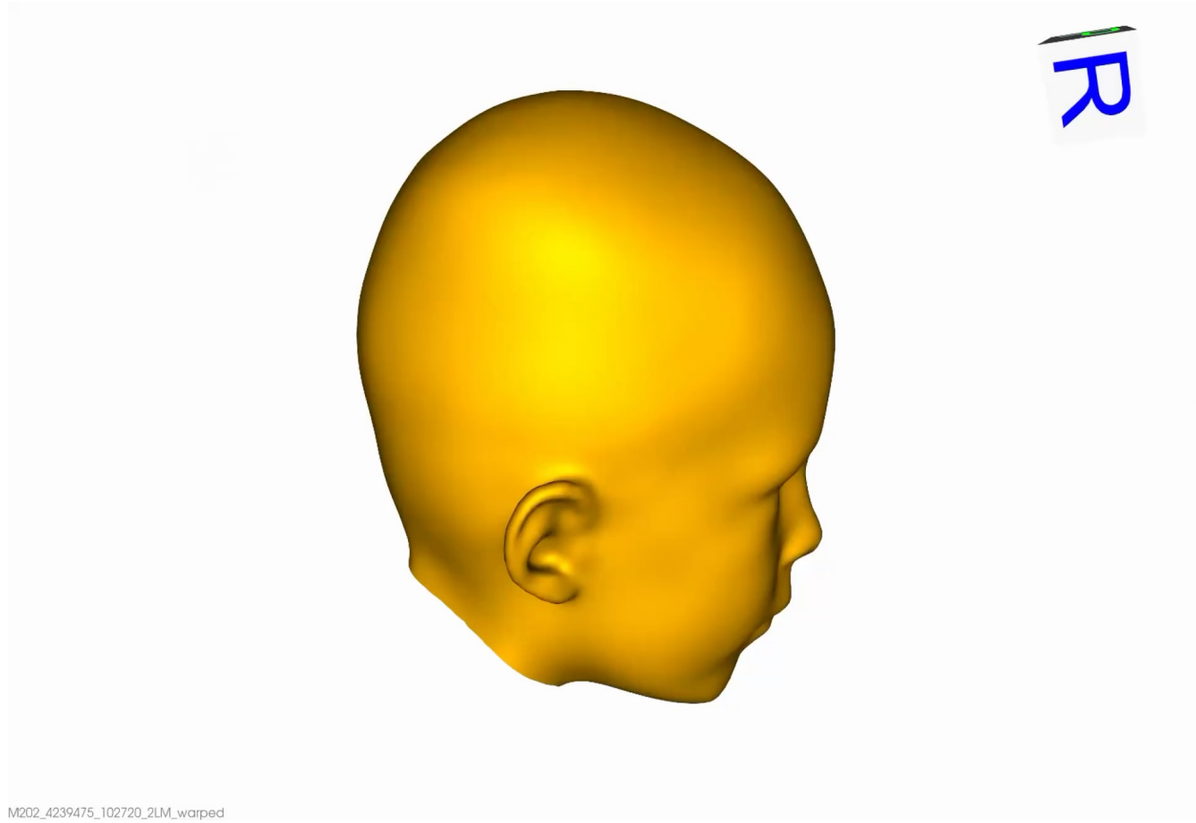
✗ Missing regions (occlusions, hair, motion)

✗ No surface correspondence

Without correspondence, 3D shape cannot be compared—so severity cannot be quantified

CranioRate 2.0 → Now Extending to 3D Photography





Gray = Raw 3D photogrammetry (incomplete, noisy, variable coverage)

Yellow = Standardized surface representation (missing regions imputed, non-anatomy removed)

Transforms incomplete and noisy 3D photos into consistent, analysis-ready cranial shape

Learns anatomically plausible completion from noisy, partial data

Establishes dense correspondence across patients

Moving Forward

National Institutes of Health

NIDCR - R01 FUNDING
2023-2028

EXPAND PHENOTYPES

Beyond sagittal → full craniosynostosis spectrum

LINK SHAPE TO OUTCOMES

Clinical, cognitive, and genetic data

MULTIMODAL DATA AT SCALE

CT + 3D + MRI + longitudinal cohorts

Toward population-scale, multimodal craniofacial analysis

The CranioRate™ Team

Principal Investigators

Shireen Elhabian, PhD
shireen@sci.utah.edu

Jesse Goldstein, MD, FAAP, FACS
jesse.goldstein@chp.edu

University of Utah

Ross Whitaker
Ladislav Kavan
Riddish Bhalodia
Cam Christensen
Ejay Guo
Nawazish Khan
Jake Wagoner
Wenzheng Tao
Moksha Karanam
Luke Schreiber
Alan Morris
Krithika Iyer
Jadie Adams

University of Pittsburgh

Lorrie Bowman
Erin Anstadt
Ali Ayyash
Justin Bereiger
Madeleine Bruce
Liliana Camison
Angel Dixon
Luke Dvoracek
Sarah Erpenbeck
Jennifer Fantuzzo
Annie Glenney
Philip Grosse
Jaz Irgabay
Nicolás Kass
Janina Kueper
Aditya Mittal
Joe Mocharnuk
Miles Pfaff
Tobi Somorin
Li Wang
Xiao Zhu
Tiffany Jeong
Michael Hernandez
Alzbeta Novotna

Funding

NIH/NIBIB (R21-EB026061) 2018-2021
PSF (NEPS-935975) 2022-2023
PSF RFA 2024-2025
NIH/NIDCR (R01-DE032366-01) 2023-2028

CranioRate™: A Common Language for Cranial Shape

Enables objective, multi-center benchmarking at scale

Representation enables clinical AI

Standardizes how cranial shape is measured, shared, and studied

CranioRate™

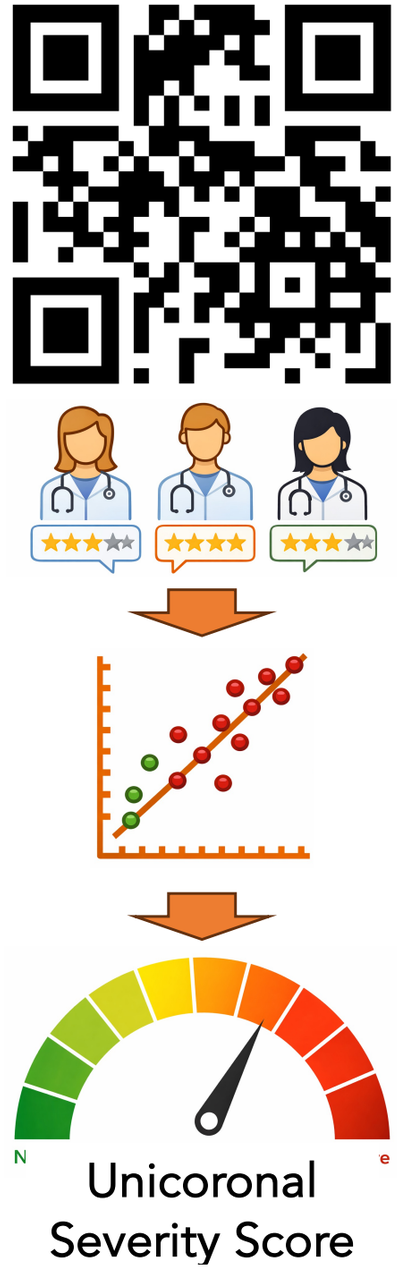
Join the CranioRate™ Consortium!

Scan to collaborate!

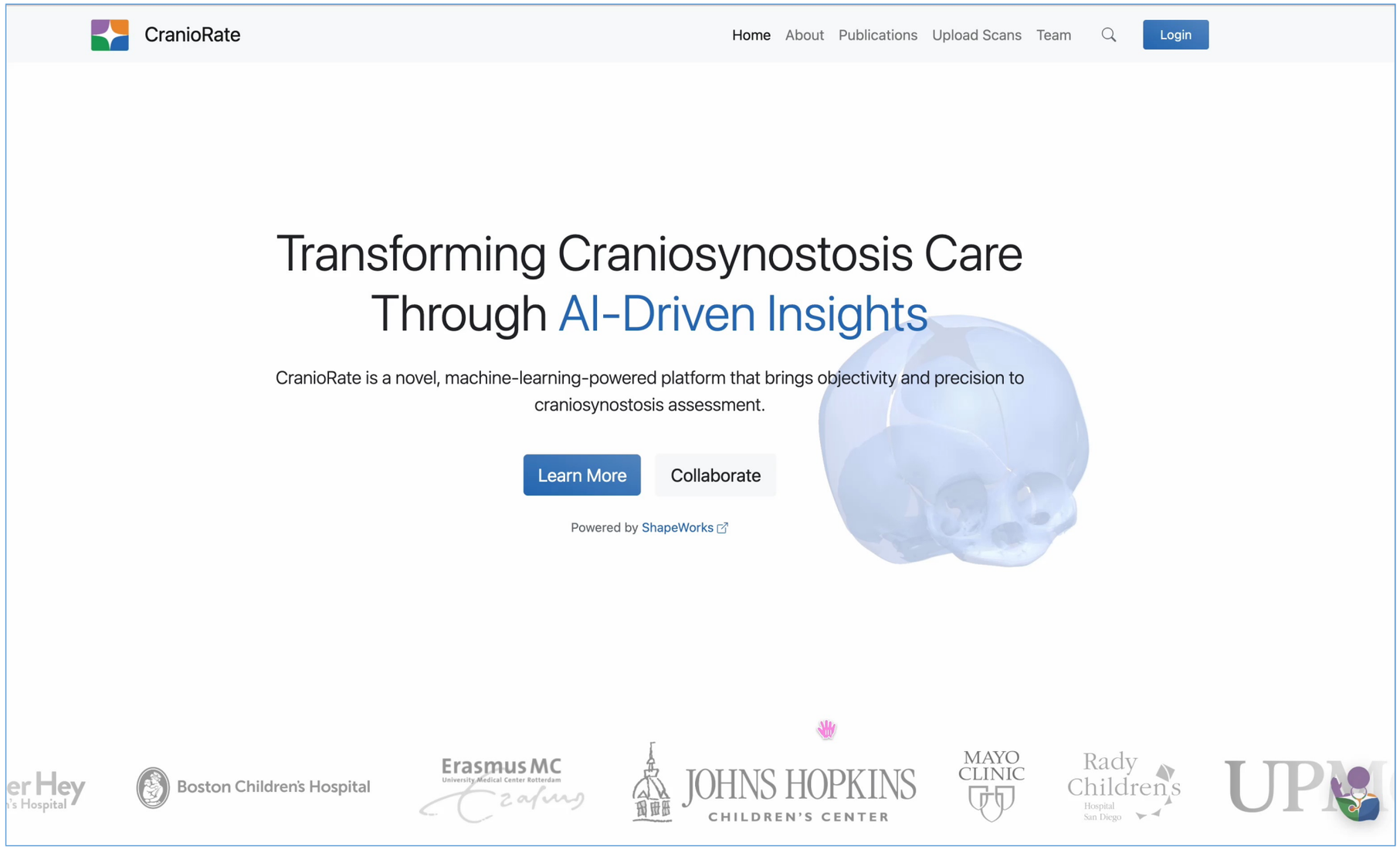
CranioRate Email: craniorate@gmail.com

Signup Form

Collecting expert ratings for unicoronal severity

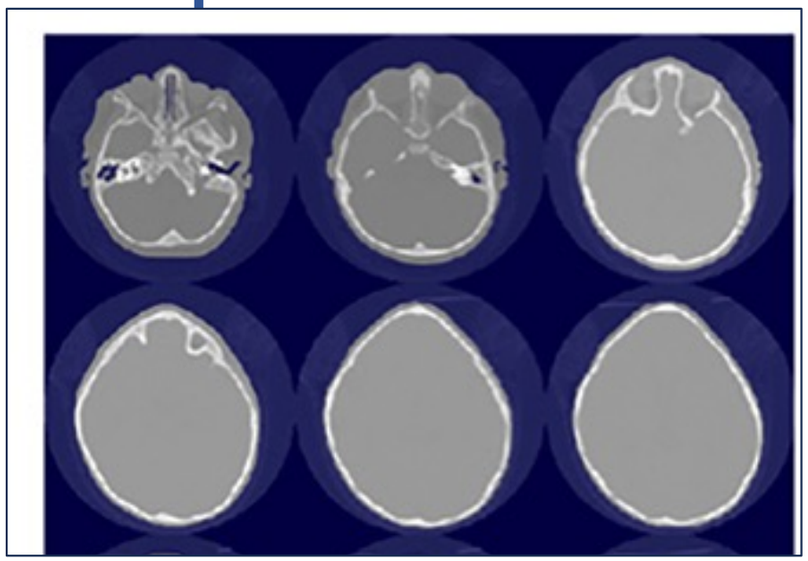


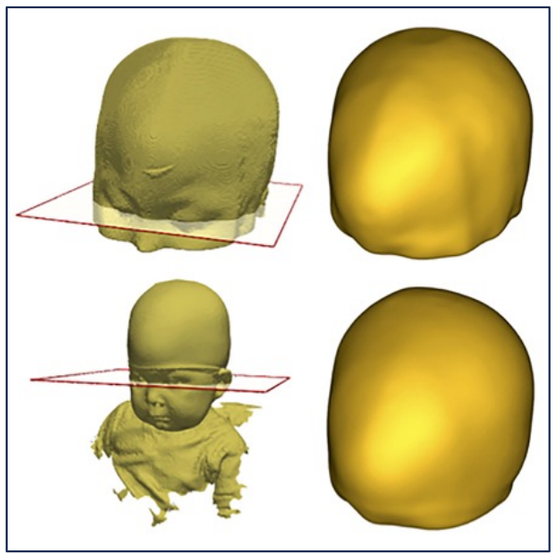
CranioRate™ at the Point of Care



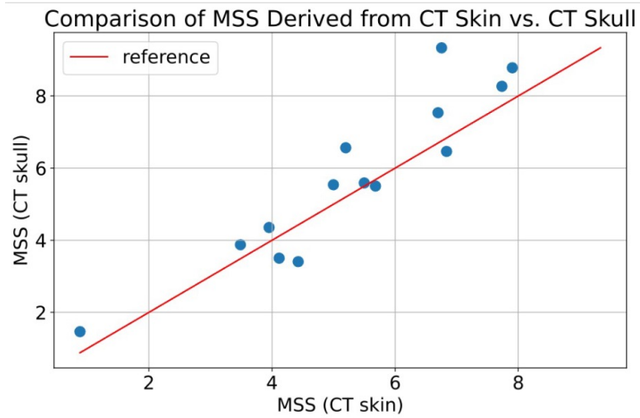
Upload a CT → Receive quantitative severity scores

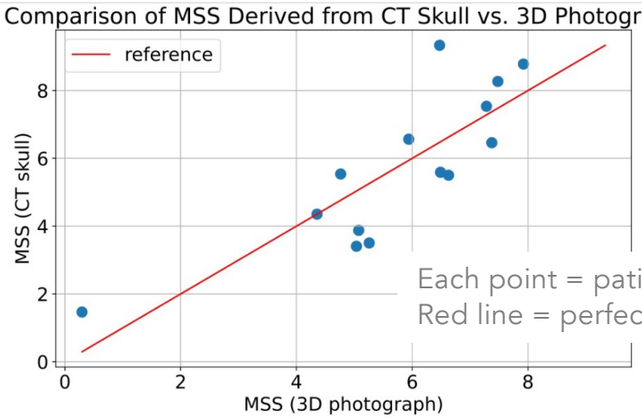
3D photos match CT-based severity scoring



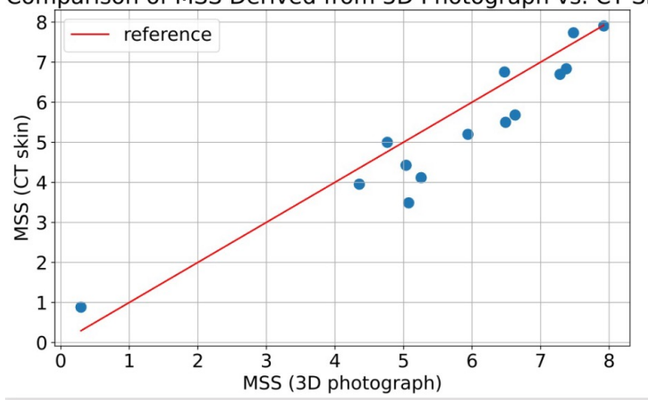


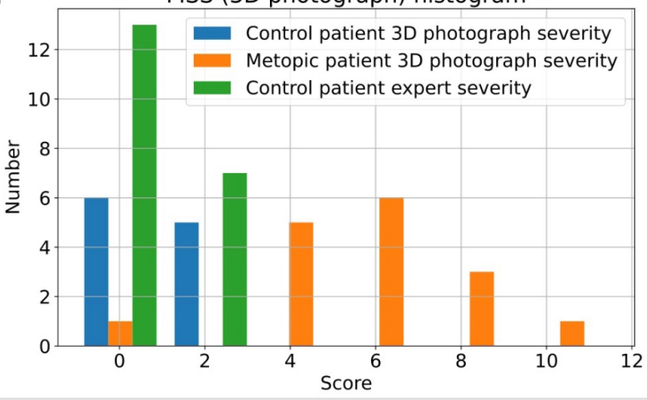
Manual processing + alignment required





Each point = patient
Red line = perfect agreement





Strong agreement with CT-derived MSS

⚠ Requires manual preprocessing → not scalable

Validates generalization across imaging modalities

(Bruce et al, CPCJ, 2023)

Original Article

3D Photography to Quantify the Severity of Metopic Craniosynostosis

The Cleft Palate Craniofacial Journal 2023, Vol. 60(8) 971–979
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DOI: 10.1177/10556562211087071
journals.sagepub.com/home/cpc

Madeleine K. Bruce, BA[1,#], Wenzheng Tao[2,#], Justin Beiriger, BSE[1], Cameron Christensen[2], Miles J. Pfaff, MD, MHS[1], Ross Whitaker, PhD[2], and Jesse A. Goldstein, MD[1]