

Breaking Barriers: A Model for Rare Disease



The Problem: Largest CCM Patient Databases in the US

Institution/Project	Total US Adult Registrants	Total Black Registrants	% Black Registrants
Angioma Alliance	914	16	1.75%
U of Chicago	512	45	8.8%
BVMC	537	2	0.4%
Mayo Clinic	282	5	1.8%
Total	2245	68	3.0%

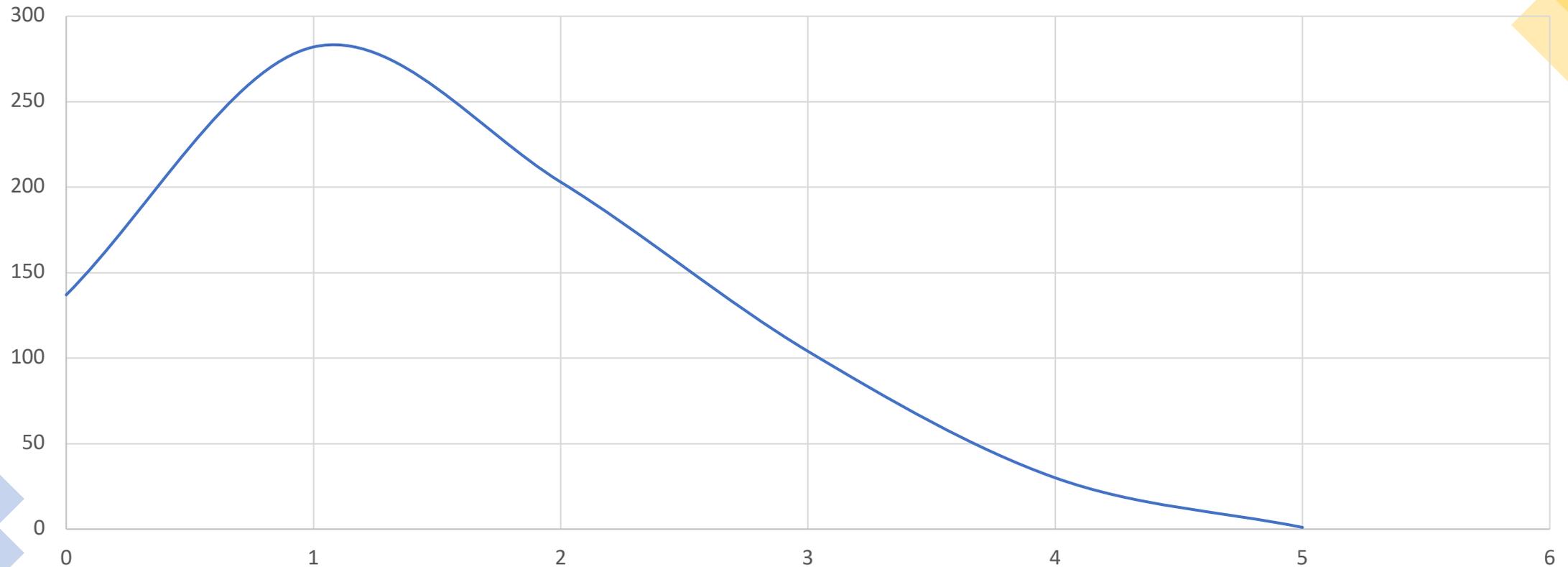
What the
numbers
should
be

- Based on epidemiology, there are approximately 80,000 Black Americans with a cavernous angioma lesion in the US
- If diagnostic rates were consistent with other races, **there should be about 20,000 Black Americans who are symptomatic and diagnosed.**
- We know 68 of these patients. The remainder who are diagnosed are missing critical information for healthcare.

Lower CCM Diagnostic Rates? A Clue: Level of Disability

White Non-Hispanic Registrants

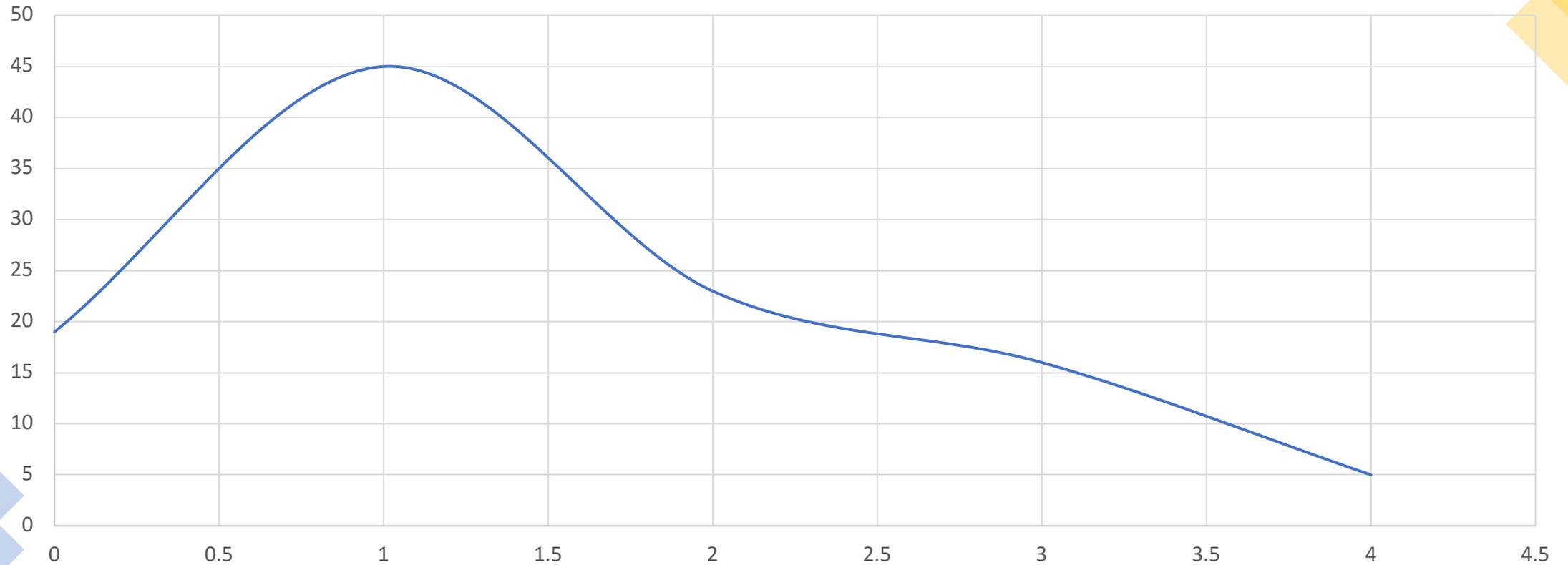
Number of Registrants at Each Level of Disability



Hispanic/Latinx Registrants

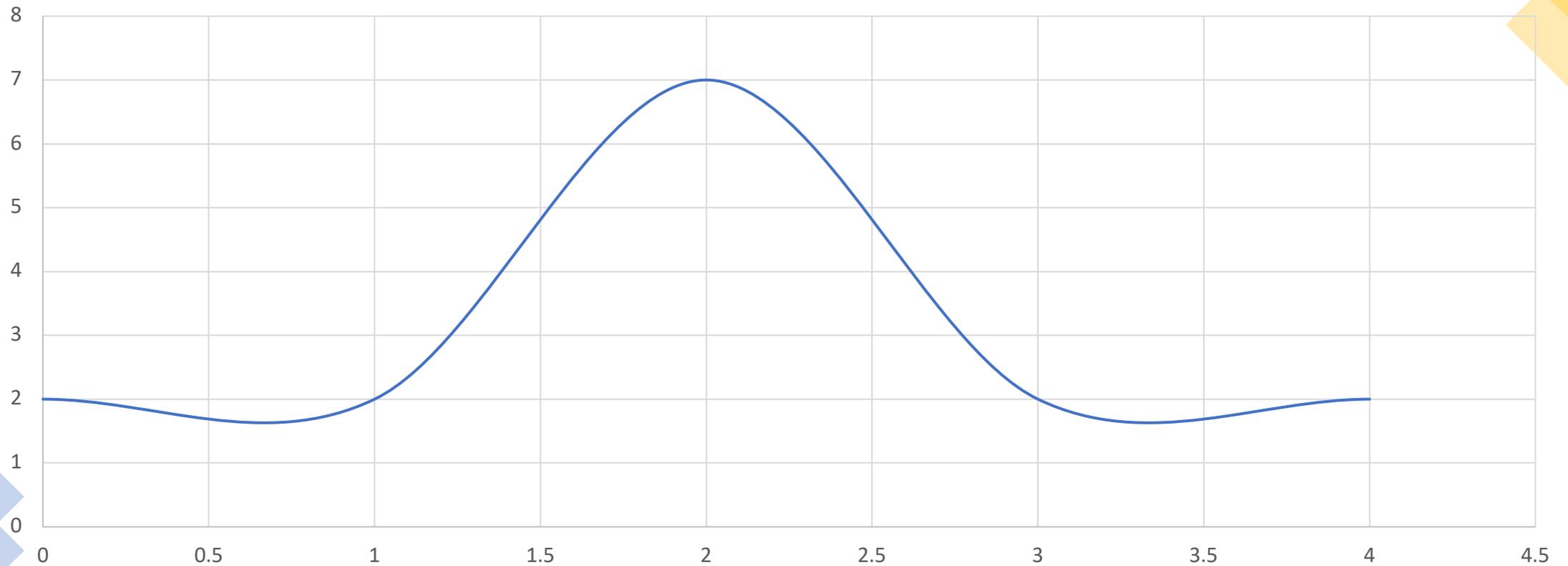
This curve is very similar to non-Hispanic Whites

of Registrants at Each Level of Disability



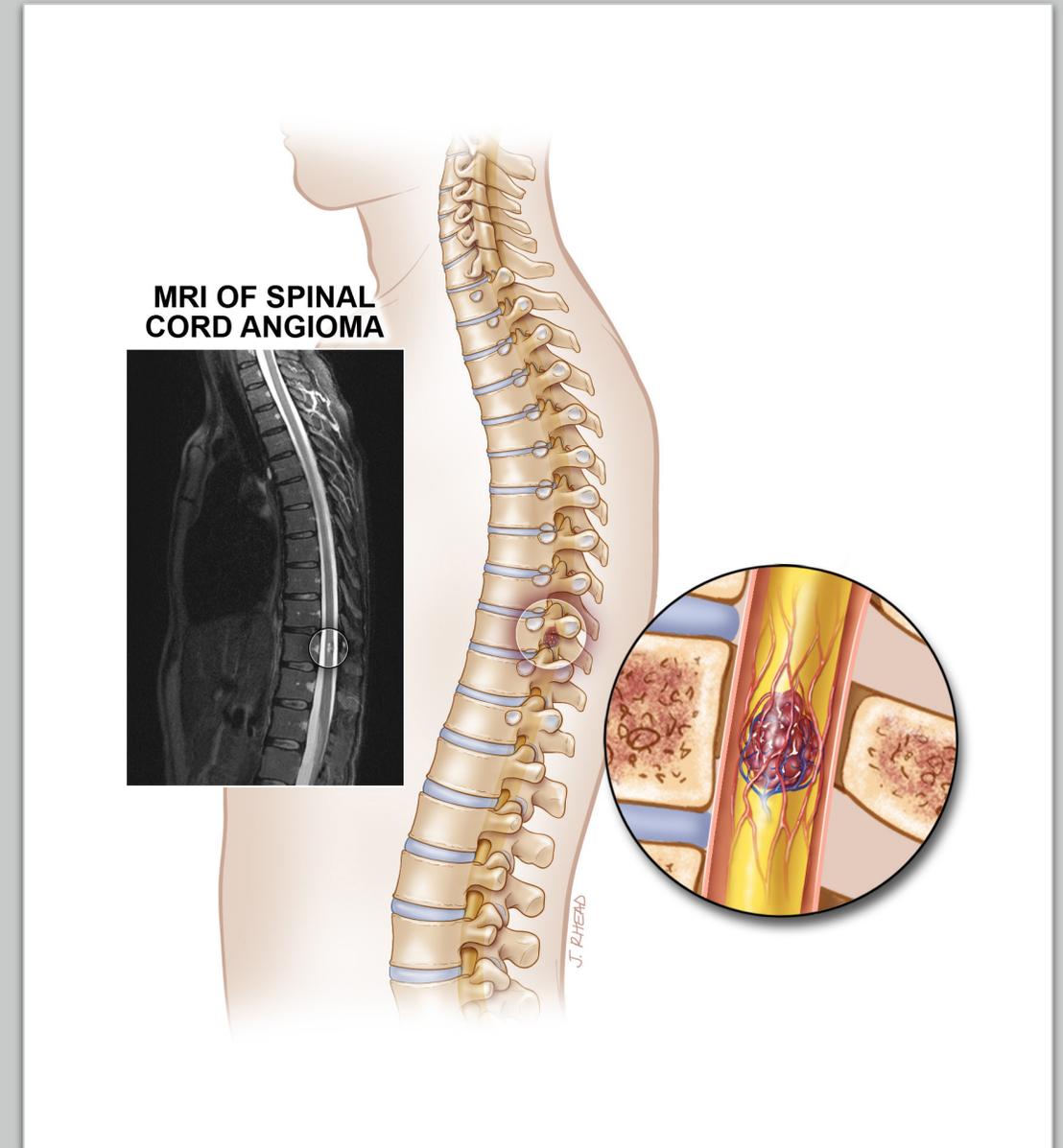
Black Registrants: The numbers are small, but there is clearly more disability

Number of Registrants at Each Level of Disability

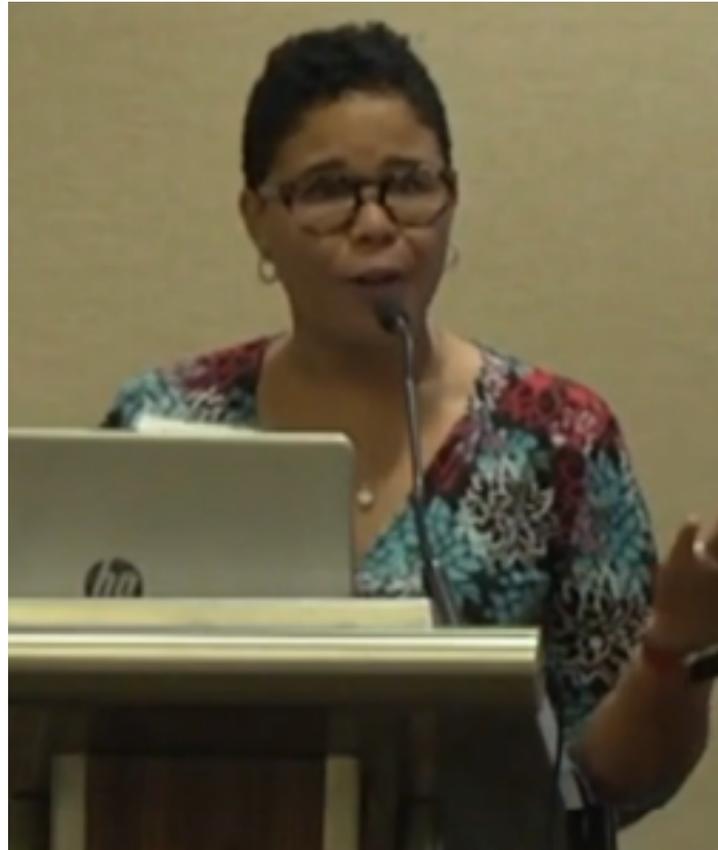


What else we know

- Black Americans in our database are much more likely to have a lesion in the **spine** (50% have a spinal lesion vs 10% in the rest of our registry)
- Spinal lesions can result in more obvious, disabling symptoms that are harder to ignore or misinterpret.



Examples of CCM Diagnosis/Treatment



Angioma Alliance Board Members

- Kandance Weems-Norris
- Dr. Kimberly Foley

This Problem Crosses Rare Diseases

- There are 7000 rare diseases, affecting 10% of Americans
- 4.3 million Black Americans have a rare disease.
- Aside from a few rare diseases, such as sickle cell and lupus that disproportionately affect Black Americans, there are **no** initiatives to find, diagnose, and engage Black Americans who have rare diseases.
- Patient groups all use a one-size-fits-all engagement approach that is not working for this community.

Project Goals

Find and Engage Patients

Find and engage diagnosed Black CCM patients, particularly to convey disease mitigation information and to foster community and conversation.

Improve Diagnosis

Improve diagnostic rates through provider education so that people come to diagnosis earlier and can benefit from known interventions.

Increase Representation in Research

Increase Black representation in research databases.

Create a Model for Rare Disease

Create an outreach and education model that can be emulated by other rare disease groups and expanded by us.

Angioma Alliance Strengths

- We have had a **successful Hispanic outreach program** in the Southwest that incorporated creative, culturally exciting ways to engage this group.
- We are the **only patient advocacy organization** for this illness, and we are viewed as a **model for other rare disease organizations**.
- The moderately successful outreach accomplished through the **University of Chicago** gives us something to build on.
- We have **connections and partnerships in many intersecting areas** – stroke, epilepsy, health equity, research, and rare disease.

Methods



LISTENING &
CONNECTING



PARTNERSHIPS



HOSPITAL
OUTREACH



DOCUMENTATION
AND MENTORSHIP

Listening & Connecting

Our first job is to listen

- We will interview patients in our registry and from the other national databases. We will create opportunities for these patients to meet each other to begin to create a community.
- We will interview other stakeholders and subject matter experts – family members, healthcare providers, community health organizations, lupus and sickle cell disease organizations, etc. to understand the need and learn best practices.

Partnership with Univ. of Virginia

- UVA will be serving as an additional source of data. They will characterize their CCM patients (approximately 500) to help us better understand any between-group differences.
- UVA is a public hospital, with a multi-state patient-base. At first glance, the proportion of patients diagnosed with CCM at UVA who are Black is proportional to the surrounding community. We will be digging deeper into this data.
- We are currently awaiting UVA IRB review and approval of the project, with Dr. Abimbola Sunmonu, Vascular Neurology Fellow as PI. This work is not part of our JGF funding request but is complementary.

Anticipated Partnerships

We will create, strengthen, and leverage partnerships to extend our reach.

- Patient Engagement – Black Women’s Health Initiative, Epilepsy Foundation, American Stroke Association, YoungStroke, Minority Stroke Working Group, National Black Church Initiative & others
- Healthcare Providers - American Medical Association Center for Health Equity, the National Medical Association & others
- Research Engagement – local programs such as the Northwestern Center for Community Health
- Rare Disease – National Organization of Rare Disease, Global Genes, Genetic Alliance, and EveryLife Foundation

Public Hospital Outreach

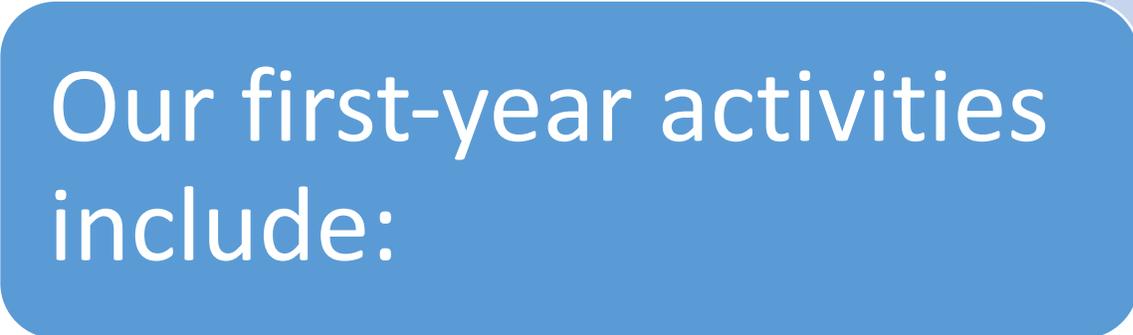
- We will engage in targeted outreach to medical providers in 3-5 large public hospitals. Top possibilities include hospitals in Chicago, Brooklyn, Philadelphia, Houston, Detroit, Atlanta, Memphis, Oakland, and Baltimore.
- We will support outreach efforts by our Centers of Excellence to their local public hospitals. We will offer patient materials and support as well as provider training.

Create a Model

- We will document our process and learnings.
- We will offer these to rare disease umbrella organizations and as part of mentoring other rare disease patient advocacy groups.
- We will expand our CCM model into new cities.



Project Goals



Our first-year activities include:

- Qualitative interviews
 - Connecting current patients to create community
 - Building partnerships
 - Creating materials
 - Outreach to one public hospital (pilot)
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