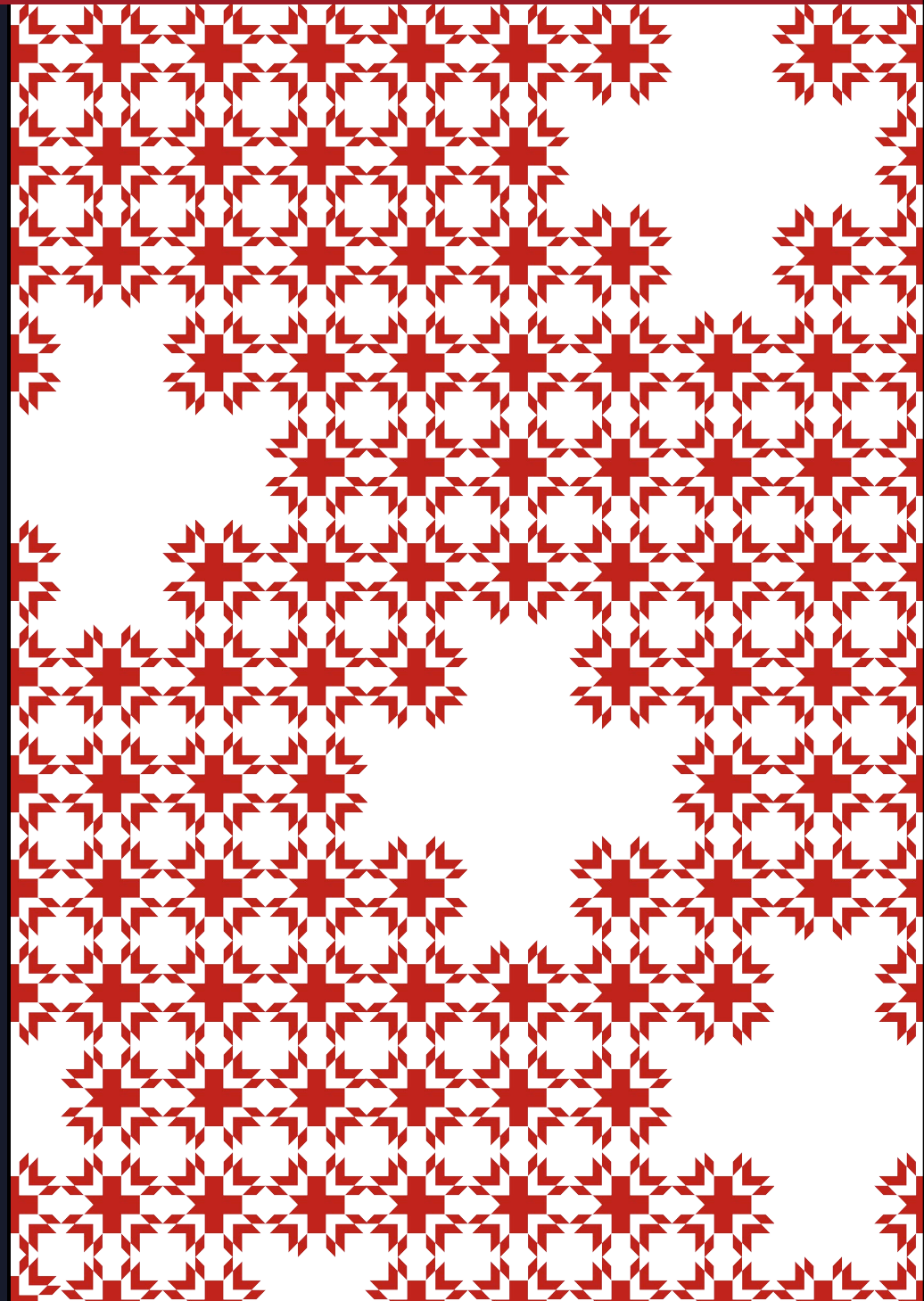


# The Challenges and Opportunities of Collaboration

Adam D. Heaps  
Laboratory Scientist



Clinic for Special Children



# Disorders @ CSC

132 known variants, 110 disorders



5 benign disorders

3 de novo dominant

2 still under review

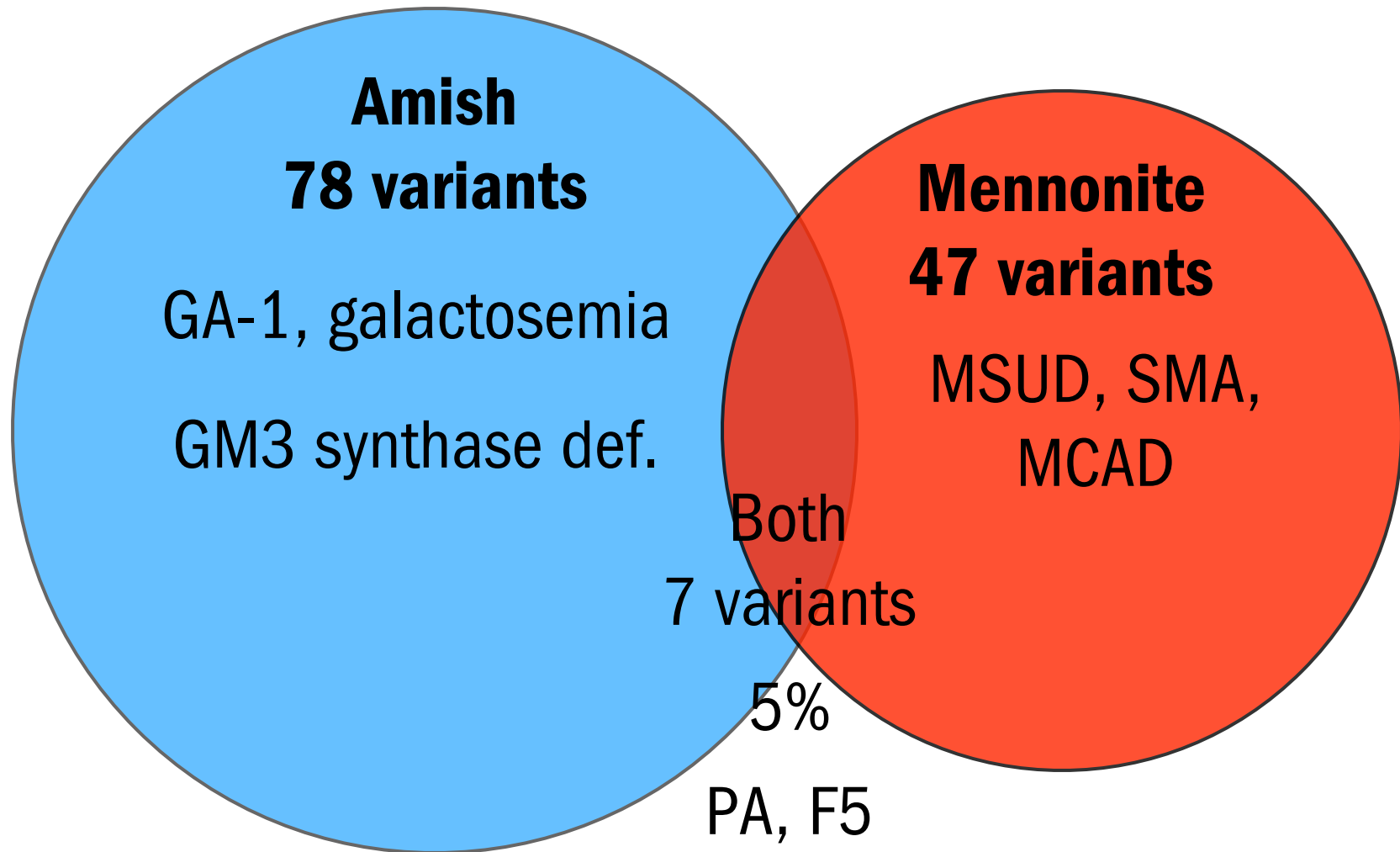


122 known variants, 100 disorders

Recessive: 107 variants, 85 disorders

Dominant: 10 ; X-linked: 5

Identify about 10 new variants/year



Same disorders, different variants: SCID, PKU

# Why Collaborate?



# CSC Weaknesses

- Functional protein studies
- Model organism work
- Staff hearing booth, cardiology equipment, ophthalmology
- Perform exome and genome sequencing
- Many biochemical assays
- Investigate each disorder in detail

# CSC Strengths

- Clinical
  - Patients with disorders, frequently serious and life threatening
    - Provide primary care, need treatments/protocols, develop and test
    - Clinical trials
    - Always something new to study
- Lab
  - Gene discovery (microarrays, gene sequencing, exome data analysis)
  - Biochemical studies (GC-MS, ELISA, HPLC)
  - Diagnostic services
- Hearing booth, cardiology equipment, ophthalmology equipment
- Understanding of the population, institutional knowledge (educate others)

# Why Collaborate?

- We can't do it all (skills, equipment, staff)
- We can't know it all
- Need input from other individuals and organizations to help strengthen a project
- New opportunities
- Advance research (patient care) in a way we can't on our own
- Fill weaknesses in our organization with strengths in another (vice versa)

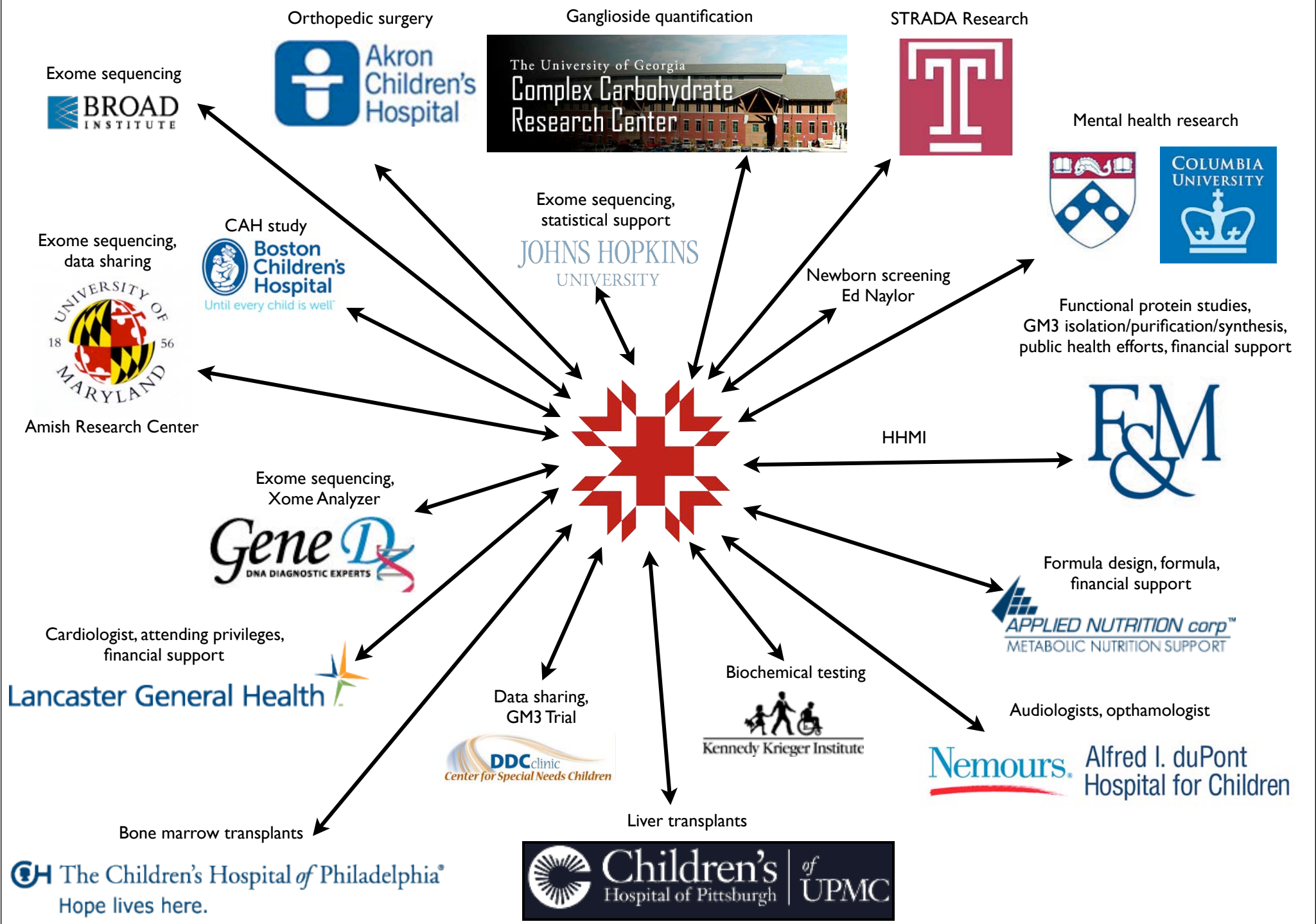
# Collaborators

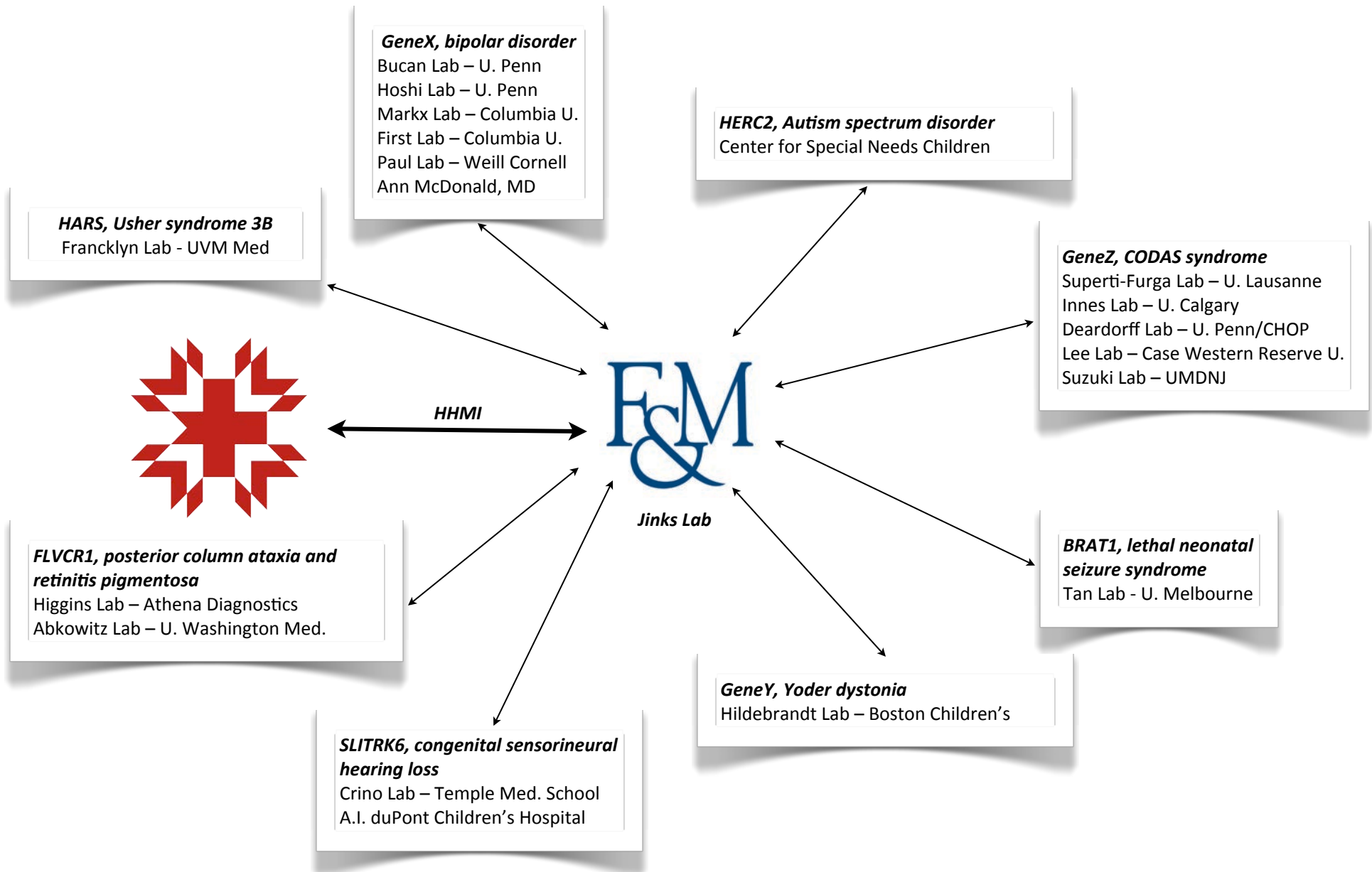
- #1: Research is patient focused
- Select collaborators carefully
  - Good working relationship
  - Communicate
- What are the manpower requirements?
- Know your limits, you can't work with everyone!



# Projects @ CSC

- CNTNAP2 - better phenotyping - oxytocin trial
- GM3 synthase deficiency - G500 trial - DDC, CCRC, Hess lab, Fonterra, Nemours
- Heredity thromboemolism - gene discovery, functional data - Jinks Lab, Amish Research Center
- Formula design (PKU, MSUD, PA) - Applied Nutrition
- Ion Torrent PGM - CSC panel validation
- Hearing loss - Nemours
- Various patient specific projects





# Franklin & Marshall

## College HHMI - Brigatti

- Unique student opportunities and education
- Funding for CSC and F&M (institutions, individual students)
  - Functional Protein Studies - Jinks
- Real world context
- Unique research and publications (student authors)
  - Public Health Initiatives - Miller, Rice, Yost
- Interaction with patients
- Good way to publicize work
- Unique educational opportunities, viewpoint
- Research into CSC disorders, benefit to patients
  - GM3 synthase deficiency - Hess, Wesalo



**Thank You!**