Massachusetts General Hospital
Founding Member, Mass General Brigham

Launching a Multidisciplinary Clinic

At Massachusetts General Hospital

July 2025

Overview

- Who We Are
- Recognizing the Need
- Benefits of Multidisciplinary Clinics
- Our Experience at Mass General
- The Vision
- Progress
- Next Steps

Who We Are

Mandy Nagy, MD

Pediatric Neurogeneticist

Mass General Brigham

Leah Wibecan, MD

Pediatric Neurologist

Mass General Brigham

Jayme Betts

Parent of Sophia, age 6

Co-Founder of Standing Tall for Sotos Initiative

Kim Little

Parent of Ellyn, age 34

Co-Founder of Standing Tall for Sotos Initiative

Clinic Faculty

Mandy Nagy, MD

Dr. Nagy received her medical degree from Case Western Reserve University and trained at MGH as a resident in Pediatric Neurology followed by a fellowship in Neurogenetics and Gene Therapy before joining the Mass General/Harvard Medical School faculty.

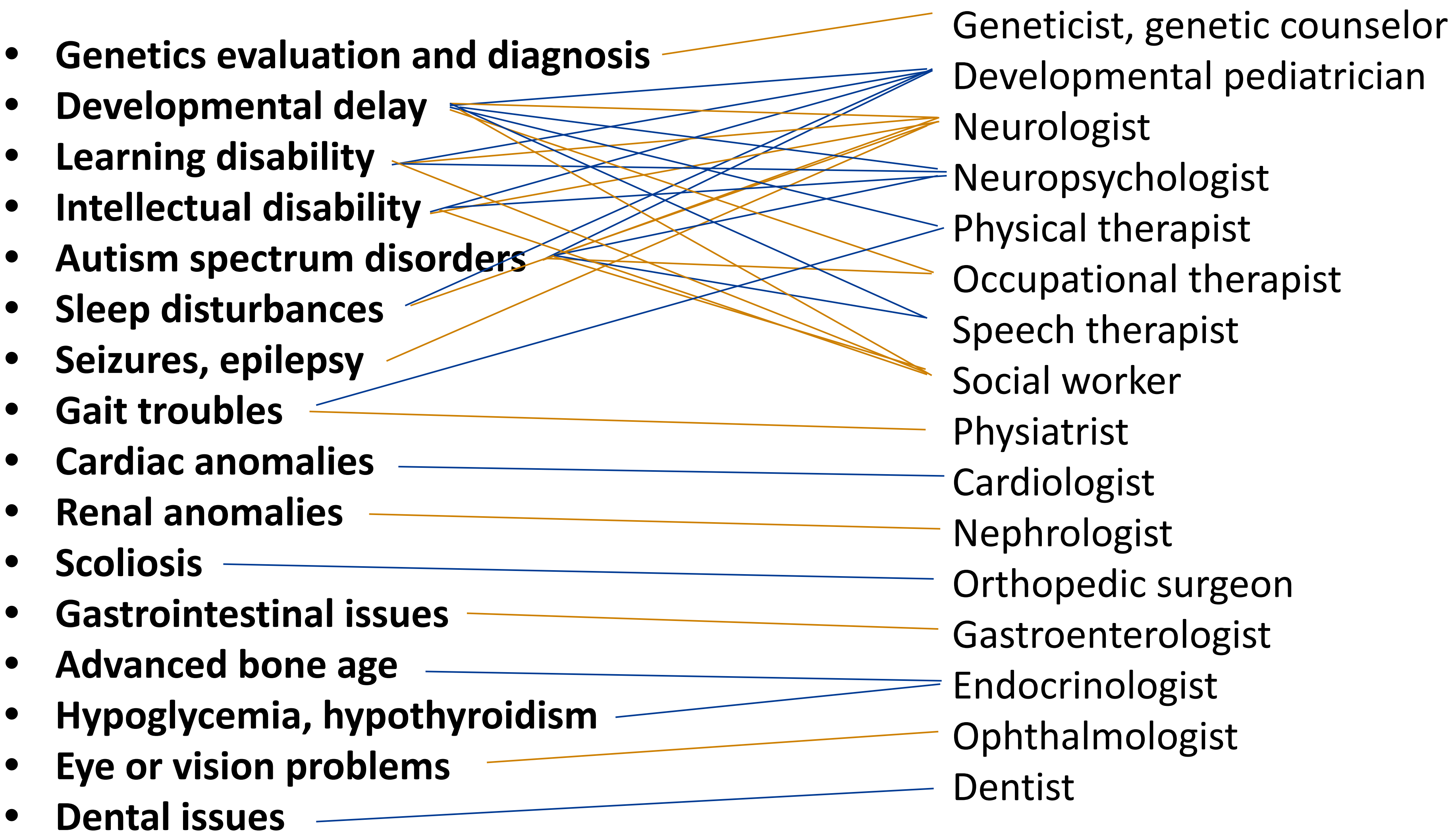
Leah Wibecan, MD

Dr. Wibecan is a pediatric neurologist at Mass General specializing in epilepsy and autism. She received her medical degree from Harvard Medical School, and completed her Pediatric Neurology residency and epilepsy fellowship at Mass General. She has an interest in strengthening care for underserved communities and in global health.

Recognizing the Need

Complex Care Needs in Sotos Syndrome

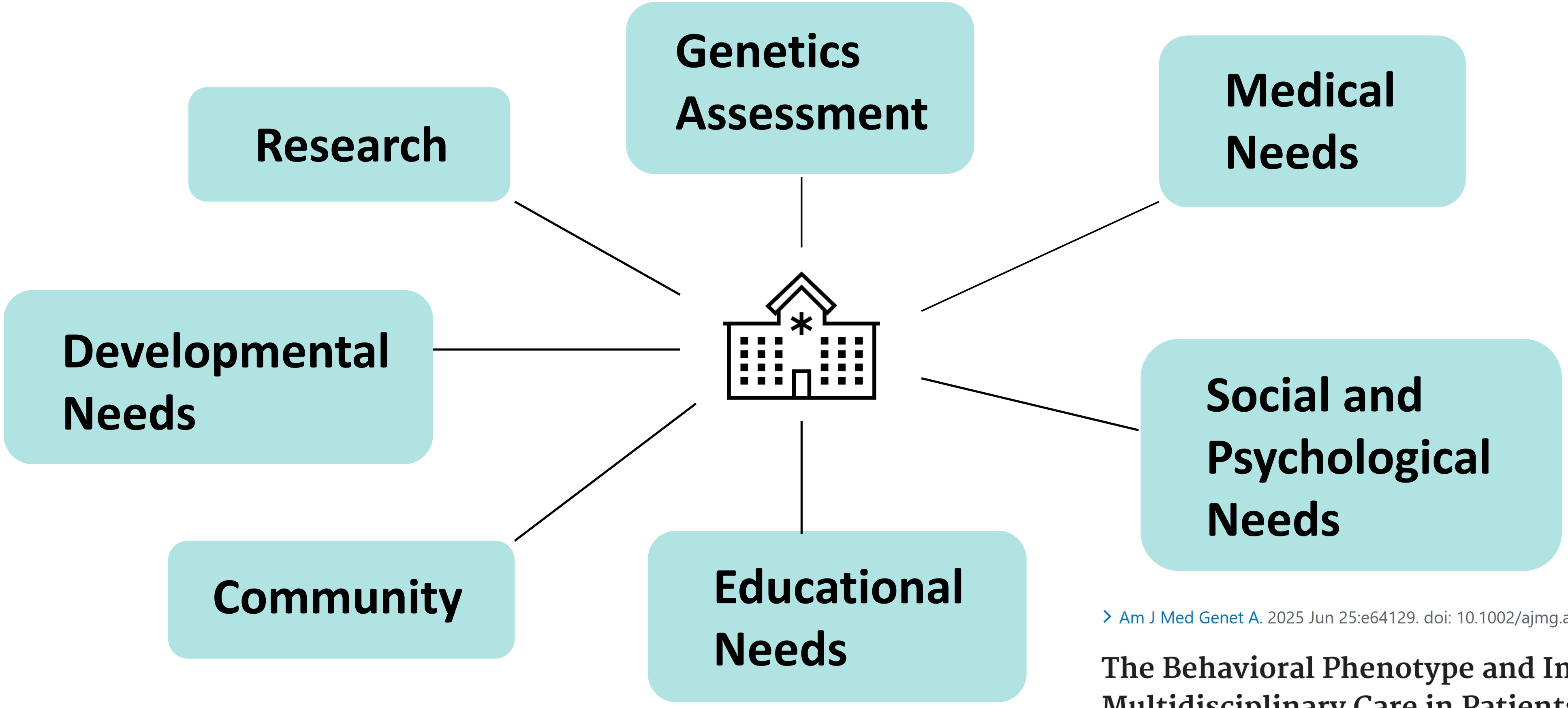
It takes a team…



- **Genetics evaluation and diagnosis**
- **Developmental delay**
- **Learning disability**
- **Intellectual disability**
- **Autism spectrum disorders**
- **Sleep disturbances**
- **Seizures, epilepsy**
- **Gait troubles**
- **Cardiac anomalies**
- **Renal anomalies**
- **Scoliosis**
- **Gastrointestinal issues**
- **Advanced bone age**
- **Hypoglycemia, hypothyroidism**
- **Eye or vision problems**
- **Dental issues**

Geneticist, genetic counselor
Developmental pediatrician
Neurologist
Neuropsychologist
Physical therapist
Occupational therapist
Speech therapist
Social worker
Physiatrist
Cardiologist
Nephrologist
Orthopedic surgeon
Gastroenterologist
Endocrinologist
Ophthalmologist
Dentist

Multidisciplinary Specialty Clinics in Genetic Syndromes



- Genetics Assessment
- Medical Needs
- Social and Psychological Needs
- Educational Needs
- Community
- Developmental Needs
- Research

> Am J Med Genet A. 2025 Jun 25:e64129. doi: 10.1002/ajmg.a.64129. Online ahead of print.

The Behavioral Phenotype and Importance of Multidisciplinary Care in Patients With Sotos Syndrome: A Single-Center Experience

Aravind Viswanathan [1], Andrew M George [1], Evan R Hathaway [1], Carlyn Glatts [2], Jennifer M Kalish [1] [3] [4]

Multidisciplinary Clinics for Genetic Syndromes

- **Tuberous Sclerosis Complex (TSC)**
  - Genetic disorder that causes benign tumors and other abnormalities in many organ systems
  - Research has shown that a multidisciplinary approach helped with:
    - Diagnosis of complex and rare multisystemic complications
    - Transition from child to adult care
    - Ensuring adherence to up-to-date best practices

- Three-step roadmap to organizing care:

1. Begin organizing care
2. Establish a core team
3. Coordinate multidisciplinary team

- Neurologist
- Neurosurgeon
- Nephrologist
- Urologist
- Pulmonologist
- Ophthalmologist
- Cardiologist
- Dermatologist
- Geneticist
- Psychiatrist/ psychologist
- Case manager for care coordination

Auvin S, Bissler JJ, Cottin V , et al. A step-wise approach for establishing a multidisciplinary team for the management of tuberous sclerosis complex: a Delphi consensus report. *Orphanet J Rare Dis.* 2019 Apr 30;14(1):91.

Multidisciplinary Clinics for Genetic Syndromes

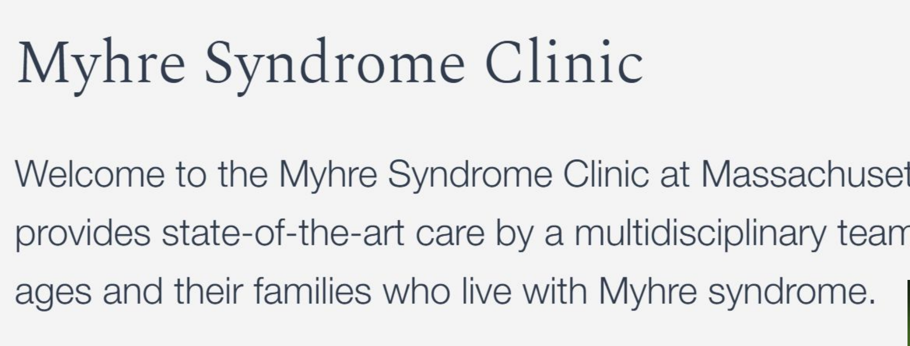
- **Key components in a TSC multidisciplinary clinic**
  - Patient care coordinator
  - List of associated health professionals
  - Organized care pathway
  - Following accepted TSC guidelines
  - Network of local or regional providers beyond the clinic
  - Patient database for research and case management
  - Plan for transition from pediatric to adult care
  - Link to family TSC organizations

Herscot Center for Tuberous Sclerosis

The Carol and James Herscot Center for Tuberous Sclerosis Complex raises awareness and provides education for people living with Tuberous Sclerosis Complex, their families, providers and others.

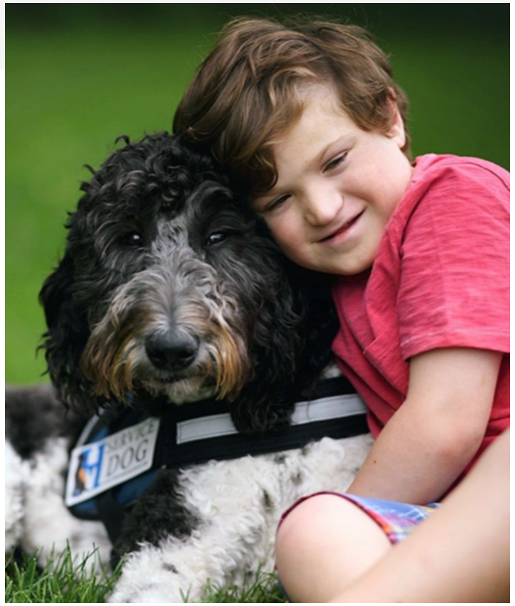
Multidisciplinary Clinics for Genetic Syndromes

- Genetic short-stature syndrome affecting cardiovascular system, lungs, musculoskeletal system, and development



Myhre Syndrome Clinic

Welcome to the Myhre Syndrome Clinic at Massachusetts General Hospital. Our clinic provides state-of-the-art care by a multidisciplinary team of providers for people of all ages and their families who live with Myhre syndrome.



- Multidisciplinary clinic followed 47 patients, identified clinical features and developed care and monitoring recommendations across all body systems

- “Medical home” with multiple specialists is able to support local providers and follow natural history over the lifespan

Multidisciplinary Clinics for Genetic Syndromes

SYNGAP RESEARCH FUND

Collaboration. Transparency. Urgency.

Children's Hospital of Philadelphia®

PROJECT 8P

Multidisciplinary Neurogenetic Clinic

CHILDREN'S HOSPITAL COLORADO

PCRC

PRISMS Clinic and Research Consortium

Smith-Magenis Syndrome Consortium Clinics

Consortium clinic locations in the US

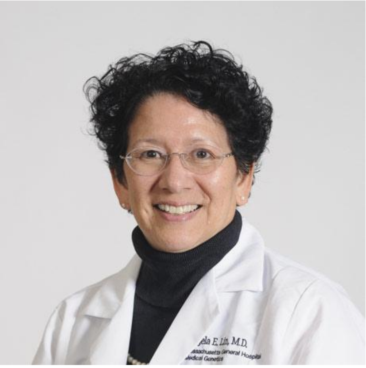
Neurofibromatosis Clinic

Massachusetts General Hospital

Founding Member, Mass General Brigham

Building the Team at Mass General

- For the past 20 years, Dr. Angela E. Lin has cared for people of all ages with Sotos syndrome
- Involvement in the Sotos Syndrome support group community in New England and nationally
- Investigation into the causes of Sotos syndrome and publication of the findings
- Collaboration with physicians and researchers nationally
- Reviewing the neuroradiological findings in Sotos syndrome
- Launched and operates two other successful dedicated clinics at Mass General, for Myhre and Turner Syndromes
- Established core team for Sotos Syndrome Clinic, with Dr. Wibecan (pediatric neurology) and Dr. Nagy (neurogenetics)



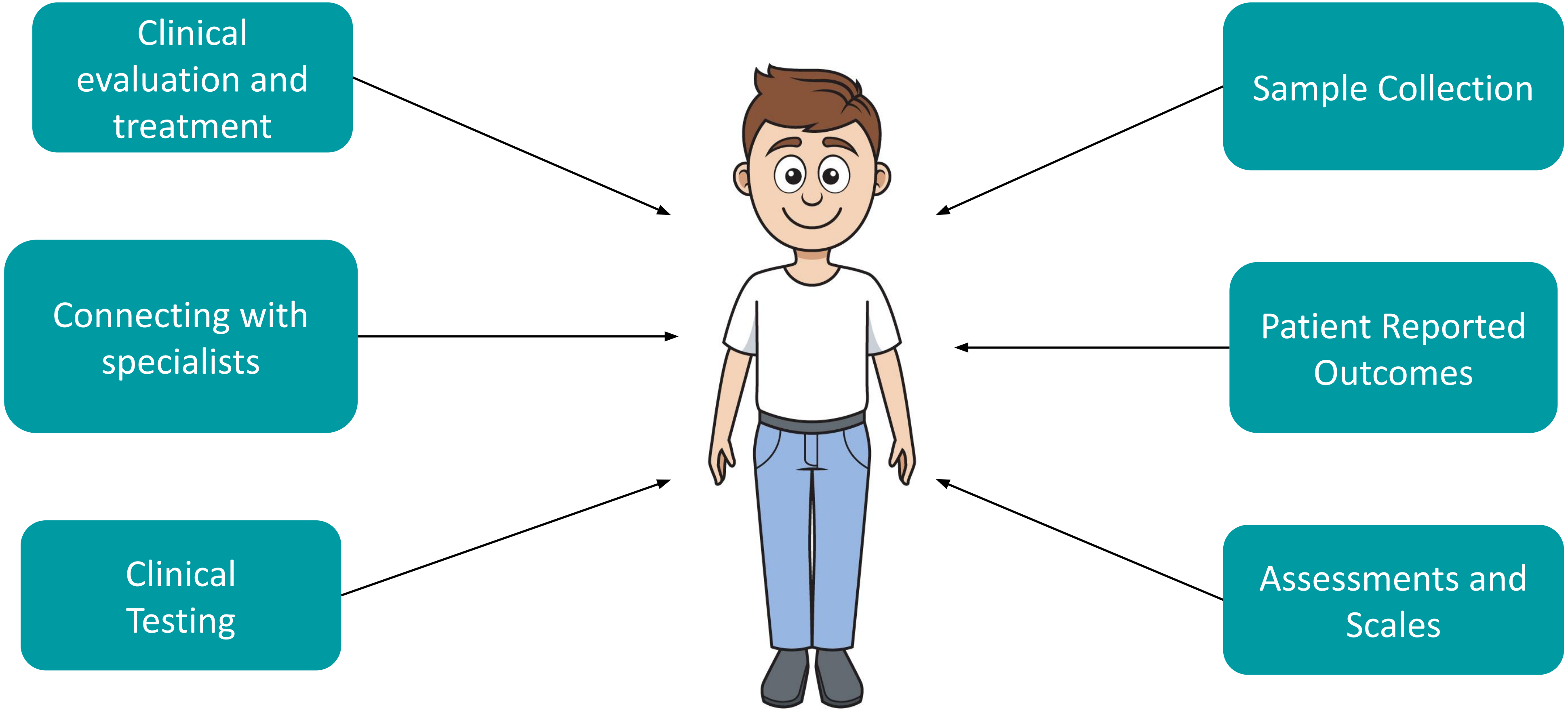
Angela Lin, MD

Geneticist

Mass General

**** Dr. Lin's Publications:***
The Neuroimaging Findings in Sotos Syndrome" (American Journal of Medical Genetics, 1997)
"Exclusion of Growth Factor Gene Mutations as a Common Cause of Sotos Syndrome" (American Journal of Medical Genetics, 2001)
"Adults With Sotos Syndrome: Review of 21 Adults With Molecularly Confirmed NSD1 Alterations, Including a Detailed Case Report of the Oldest Person" (American Journal of Medical Genetics, 2011)

Integrating Clinical Care and Research



Clinical evaluation and treatment

Connecting with specialists

Clinical Testing

Sample Collection

Patient Reported Outcomes

Assessments and Scales

Benefits of a Patient Database

Data Patterns: A comprehensive database helps identify common symptoms and early signs.

Understanding Treatment Efficacy: With patient data, we can analyze how different patients respond to treatments.

Longitudinal Data: Track disease progression over time, uncovering new insights into the disorder.

Biomarkers: Standardized sample collection allows for the discovery of biomarkers

Clinical Trials: Facilitates trial implementation and enrollment

Collaboration: Facilitates cooperation across research institutions, enhancing the quality of studies.

Patient Advocacy: A well-documented patient base strengthens advocacy efforts and raises awareness about the disorder.

The Vision

- Centralized care for patients
- Access to a wide range of specialists working together to evaluate and treat
  - *Clinicians, Social Worker, Nurse, Clinic Coordinators, Clinical Research Coordinator*
- Simplify the complexities Sotos families face after diagnosis
  - *Creating a care guide resource for newly diagnosed families*
- Familiarity with navigating and coordinating the educational and psychosocial needs of Sotos patients throughout their life
- Data collection to understand the prevalence of various conditions associated with Sotos Syndrome, made available to Sotos community and medical professionals.
- A research program focused on Sotos Syndrome, including clinical trial initiatives to advance understanding and treatment options for patients
- Raising awareness through educational initiatives and trainings

Progress

1

Collaboration

2

Vision

3

Raising Seed Funding

4

Opening Clinic

5

Sustainability

Ongoing Work

- Raising awareness among medical trainees hosted on February 18, 2025
- Developing a Sotos patient survey
- Building out registry/data collection questionnaire
- Determine the research that our community want to pursue
- Work with international community to establish a registry
    - Coordination of Rare Diseases at Sanford (CoRDS)
    - Based at Sanford Research, a nonprofit research institution, CoRDS is a centralized international patient registry for all rare diseases.
- Launching a clinic website
- Continuing fundraising and raising awareness: because.MassGeneral.org/StandingTallforSotos

Please take our survey!

- What would you be interested in seeing in a specialty clinic for Sotos Syndrome?
- Which services/specialties would be the most impactful to have represented in a Sotos Syndrome Clinic?
- If a care guide was developed for Sotos Syndrome, what issues would you most like to be addressed?
- What are some questions you had when your child was newly diagnosed with Sotos Syndrome, where it was difficult for you to find answers?
- What research priorities are most important to you?

Thank You!