



Biochemical Genetics Laboratory



Left to Right: Haoyue Zhang, James Beasley, Scott Chasse, Nadia Jakoub, Rejina Khahka, Stephanie Terry, Iskren Menkovic, Ashlee Stiles, and Patty McCaw

DESIGN BY: MEGHAN REID

Leadership and Team

Director(s): Sarah Young Ph.D., FACMG;
Deeksha Bali Ph.D, FACMG;
Ashlee Stiles Ph.D., FACMG;
Dwight Koeberl, M.D., Ph.D., FACMG
*FACMG: Fellow of the American College of
Medical Genetics

Manager: Scott Chasse Ph.D.

Admin: Kiah Rivers

Number of lab team members: 15

Focus and Volume

Lab Focus:

High complexity lab-developed tests (LDTs) for rare inborn errors of metabolism, using mass spectrometry and enzymology.

Average yearly volume:

20,000 tests



Prep Lab



Left to Right: Dr. Iskren Menkovic, Dr. Sarah Young, Dr. Deeksha Bali, Rejina Khadka, Nadia Jakoub and James Beasley.

Location and Hours

Location:

801-6 Capitola Drive
Durham, NC 27713

Hours: 6:30AM- 6:30PM



Fun Facts

Approximately 40 years ago, clinicians and scientists in the Department of Pediatrics, Division of Medical Genetics, were among the first groups to introduce tandem mass spectrometry into the clinical lab (The Mass Spec Facility). David Millington, Ph.D., Charlie Roe, M.D., Ph.D., and others in the team invented the expanded newborn screening assay for acylcarnitines and amino acids. This test is now used around the world to screen millions of babies for treatable inborn errors of metabolism.

Around the same time, Dr. Y-T Chen established an enzyme testing lab for the glycogen storage diseases (The GSD Lab). These two labs were combined to create the Duke Biochemical Genetics lab.

40 years on, we are proud to continue the legacy of our predecessors of innovating new tests and providing an essential service for the diagnosis and management of patients with rare genetic diseases.



Mass Spec Room

New in the Lab

A small change is that we are welcoming back hydroxychloroquine testing!

Our big news is that we will be moving back on campus in 2025 and combining forces with the DCAL mass spectrometry group to form the Mass Spectrometry Center of Excellence.

What does BCG do?

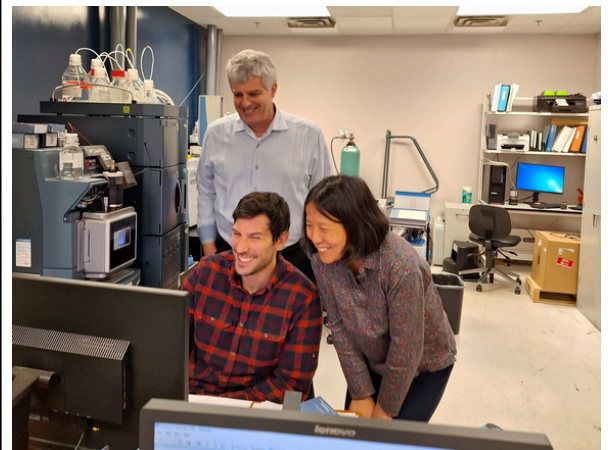
The DUHS Biochemical Genetics Laboratory is comprised of two closely integrated sections, the **Mass Spectrometry Laboratory** and the **GSD/LSD Enzymology Laboratory**.

The Duke Mass Spectrometry Section offers core metabolic (urine organic acids, acylcarnitines, amino acids, and plasma carnitine) and targeted analyte testing for the diagnosis and monitoring of patients with inborn errors of metabolism. Using state-of-the-art technology, the Mass Spectrometry Section has been the site of multiple new innovations in the diagnosis and monitoring of patients with metabolic disorders.

The GSD/LSD Enzymology Section specializes in the enzymatic diagnosis of disorders of glycogen storage, lysosomal storage, and fructose metabolism utilizing a variety of laboratory methods, including tissue enzyme assays and chemical analysis of glycogen.

Taken from

<https://clinlabs.duke.edu/biochemical-genetics>



Iskren, Scott and Hayoue