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UNIVERSITY
SCHOOL OF
MEDICINE

Medical Genetics and Genomics: The Emory Experience

Michael J. Gambello, MD, PhD
Professor and Program Director
Department of Human Genetics
Emory University School of Medicine

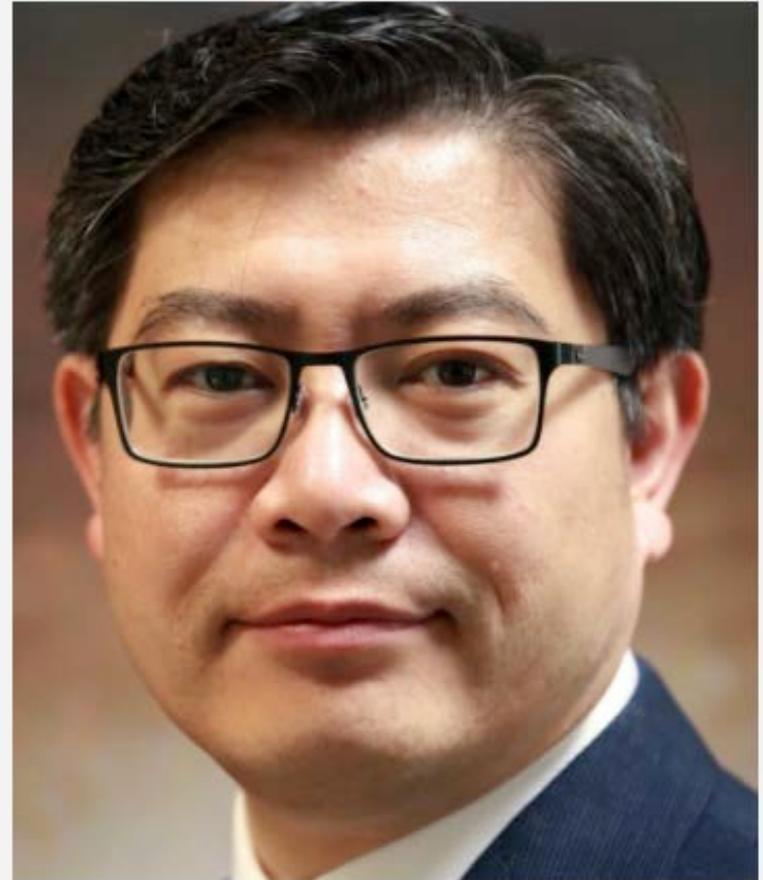


Department of Human Genetics

Welcome to the Department of Human Genetics (DOHG). It is an exciting time for human genetics with the ongoing revolution in genetics and genomics. Our unique combination of a full-fledged basic research faculty along with the comprehensive clinical genetics division places us at the forefront of contemporary translational research and predictive, precision health.

[Peng Jin, PhD](#)

Chair, Department of Human Genetics



History of the Division of Medical Genetics in the Department of Human Genetics



1970 Founded by
Louis "Skip" Elsas II, M.D.
in the Department
of Pediatrics

"Skip" Elsas II, M.D



2001-2020 Former Chair,
Stephen T. Warren, Ph.D.
founded
The Department of
Human Genetics

Stephen T. Warren, Ph.D.

2002 The Division becomes affiliated with The Department of Human Genetics emphasizing the important relationship between medical genetics and human genetics research.

<https://med.emory.edu/departments/human-genetics/patient-care/physicians-counselors.html>

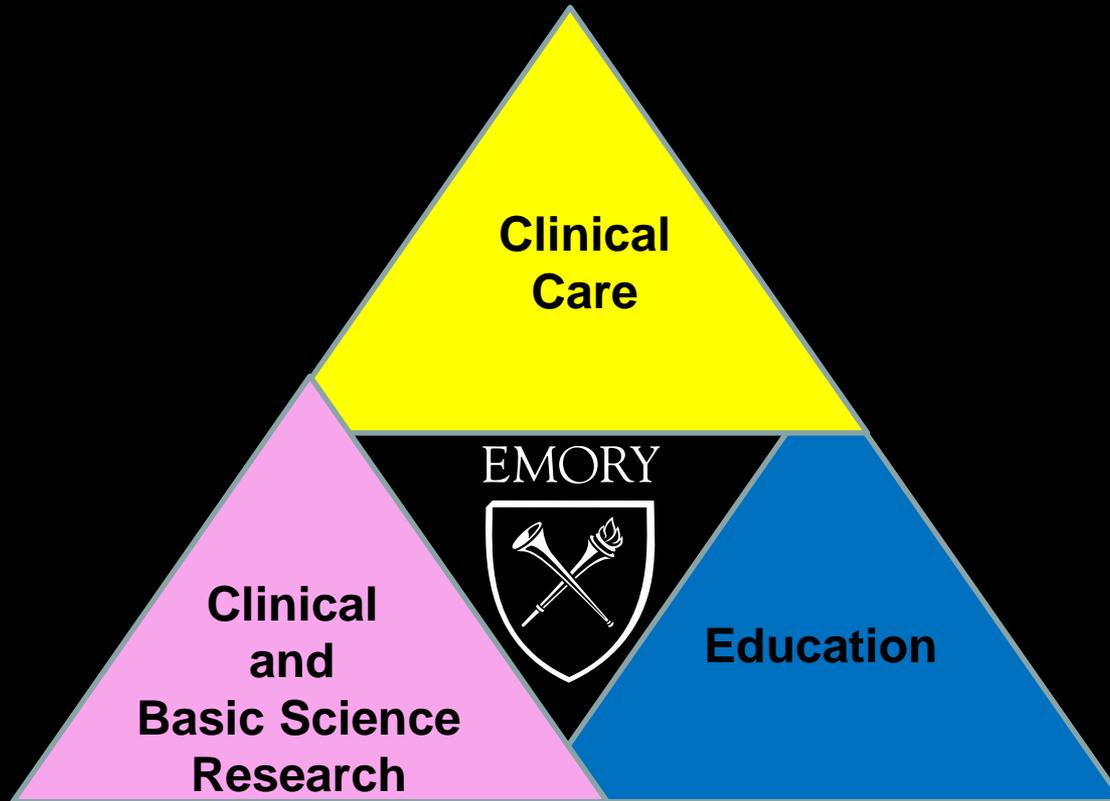


Division of Medical Genetics

A Diverse Group of Genetics Professionals



Our Missions



Educational Programs in Genetics at Emory

1. ACGME Accredited Clinical Programs -
Medical Genetics and Genomics (MGG)
Medical Biochemical Genetics
Pediatrics – MGG
2. Graduate Program
(GMB – Genetics and Molecular Biology)
4. ABGC Accredited Genetic Counseling Training Program
5. Human Genetics and Nutrition

Core Physician Faculty



Hong
Li, MD, PhD
Director Metabolic Clinic



Michael J.
Gambello, MD, PhD
Program Director



William
Wilcox, MD, PhD
Program Director MBG



Stephanie
Wechsler, MD



Jaime
Vengoechea, MD



Rossana
Sanchez, MD



Juanita
Neira, MD



Emily
Black, MD

Sample Schedule – Year 1

Months	July	August	September	October	November	December
Clinical Activity	General Genetics & Metabolism Clinics 4 days/week	General Genetics & Metabolism Clinics 4 days/week				
	One admin day/wk	One admin day/wk				
	One week Call	One week Call				
				NAMA October (Option to take Year 2)	Vacation one week	
Months	January	February	March	April	May	June
Clinical Activity	Self Directed Study	Cancer Genetics Winship Cancer Institute	General Genetics & Metabolism Clinics 4 days/week	General Genetics & Metabolism	Molecular Genetics Laboratory No Call/Clinic 2 weeks	Biochem. Laboratory No Call/Clinic 2weeks
	Call Optional			One week Call Outpatient Clinic 4 days/week		
	One week vacation	No Call	One admin day/wk		Cytogenetics Laboratory No Call/Clinic 2 weeks No Call	Vacation one week
			One week Call			Seld-Directed study one week

Outpatient Clinics

- 85% Pediatric 15% Adult
- 7000 visits/year
- Diverse patient population
- Large catchment area
- Referrals from
 - Pediatricians
 - Internists
 - Other Specialists
 - Hospital Follow-up
- Wait list 1-2 months
- Exploring Telemedicine



Georgia 10 million



Metro Atlanta 5 million



Outpatient Clinics

- General Genetics Clinics – Every Day
 - Intellectual disability, speech delay, dysmorphic features
 - Birth defects, failure to thrive, autism spectrum disorders
 - Family history of cancer, aortic dissection etc.

- Metabolism Clinics – Mondays, Tuesdays, Wednesdays
 - Abnormal newborn screens
 - Continuing Care
 - Lysosomal storage disease clinic
 - MPS, Gaucher, Fabry, Pompe etc.

Specialty Clinics

- Skeletal Disorders Clinic
- Craniofacial Clinic
- Fragile X and Down Syndrome Clinics
- Disorders of Sexual Development
- NF1/TSC Clinic
- 22q11.2 Clinic
- Prenatal Clinic
- Cancer Genetics Clinic

Inpatient Consultations



Majority at Children's Healthcare of Atlanta –

Egleston Campus 278 beds

Scottish Rite Campus 273 beds

Some adult consultations at Emory University/Tower Hospitals



When on call, resident is the first person called

No outpatient duties when on call



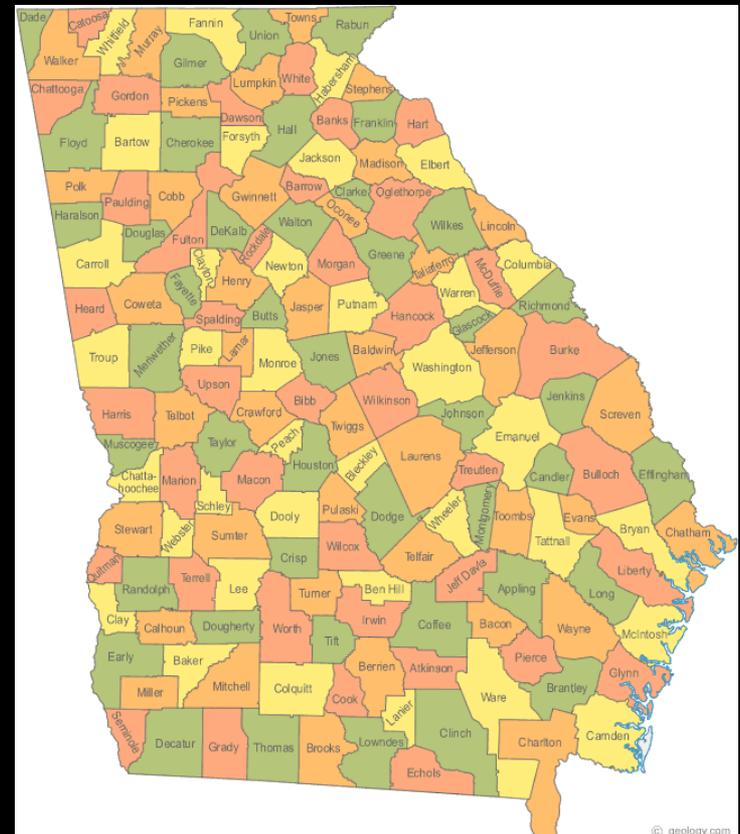
Resident always reviews plan with attending

GEORGIA Newborn Screening

Approximate Births in 2016
130,042

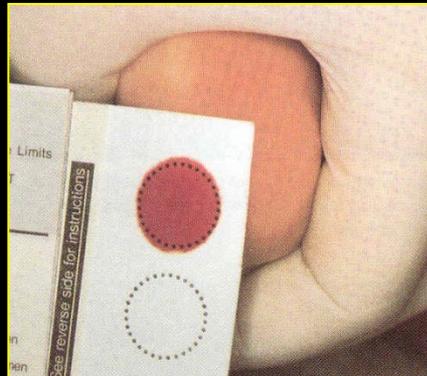
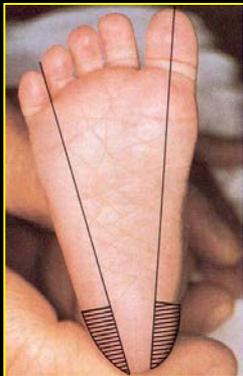
Major Racial/Ethnic Groups

- White: 59%
- American Indian: <1%
- African American: 37%
- Asian/Pacific islander: 4%
- Hispanic Ethnicity: 16% (may also be included in race categories above)



Newborn Screening Follow-up

- Division has NBS follow-up contract with GA.
- Angela Wittenauer, nurse coordinator, 6 nurses.
- NBS database
- Superb opportunity to learn about the management of positive screens for over 30 inborn errors of metabolism
- NICHD Pilot projects led by Dr. Wilcox on NBS for MPS1, Pompe disease, XALD, SMA, Homocystinuria
- Forming a Long-term Follow-up Program



Didactic Learning

- HGC 715 – Human Genetics Fall 3 credits



Kate Garber, PhD
Course Director
Multiple Lecturers

- HGC 745 – Medical Genetics Fall/Spring 3 credits



Stephanie Wechsler, MD
Course Director
Multiple Lecturers

Clinical Conference

Drs. Li/Vengoechea coordinators.

CME credits for Faculty.

Weekly one hour presentation of two cases.

Presentations by GCs, GC students, residents, medical students.

Faculty Mentor assigned. Reviews presentations and ensures content for CME.

Power Point Presentation 25-30 minutes.

Resident gives a CC once a semester

ALL VIRTUAL 2020-2021

Clinical Conference Examples from Fall 2018

Date	Presenter	Topic
09/18/18	Janette diMonda	From VUS to Diagnosis: A Case of Singleton-Merten Syndrome
	Morgan Simmons	Fabry Disease: Fact or Faux?
10/02/18	Rachel Linnemann	Diagnostic Challenges in Cystic Fibrosis
	Rhea Behlmann	
10/09/18	Emily Davis Black	Review the management of <i>PLA2G6</i> -associated neurodegeneration and explore potential therapeutic options that are in clinical trials
	Matthew Walsh	X-linked Conelia de Lange Syndrome
10/23/18	Chelsea Leonard	<i>NAA10</i> -Related Syndrome
	Georgia Loucopoulos	Patient decision-making for prenatal testing when faced with a VUS
11/06/18	Abdulrazak K.Alali	Holocarboxylase Synthetase Deficiency: A Case Presentation and Review Of The Literature
	Merlene Peter	The Tale of Two Translocations
11/13/18	Aixa Gonzalez Garcia	Inherited Systemic Hyalinosis (<i>ANTXR2</i> related disorder)
	Ellie Westfall	Challenges of <i>DICER1</i> Diagnosis
11/20/18	Christine Tallo	<i>MLH1</i> Epimutations: An Overlooked Cause of Lynch Syndrome
	Virginia Casola	A Look Into Li-Fraumeni Syndrome
12/04/18	Allison Foley	A Review of Achondroplasia and Current Investigational Treatment Options
	Tina Kushary	Clinical and Molecular Diagnosis, Screening and Management of Beckwith-Wiedemann Syndrome: A Newly Published International Consensus Statement

Clinical Conference Weekly Notice



EMORY
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THE DEPARTMENT OF
**HUMAN
GENETICS**

Continuing Medical Education
1462 Clifton Rd., NE, Suite 276
Atlanta, GA 30322
Email: cme@emory.edu
Phone: 404-727-5695
FAX: 404-727-5667

Clinical Conference January 22, 2019

Speaker

Emily Black, MD, Medical Genetics Resident PGY5

Mentor

Rossana Sánchez, MD

Title of Presentation

An Update on a Recurrent Microdeletion Syndrome

Learning Objectives:

1. Review an adult case of Smith Magenis syndrome, the clinical presentation, management and surveillance
2. Review the common deleted region in Smith Magenis syndrome
3. Discuss Birt Hogg Dube syndrome, the molecular etiology, clinical presentation and surveillance recommendations

Presenter Financial Relationships: NONE

Company/Role: Emory University
Medical Genetics Resident PGY5

Financial Relationships of RSS Director,
Planning Committee Members and Others: NONE

Speaker

Rachel Logan, MSSc, Genetic Counselor II
Children's Hospital of Atlanta

Mentor

Rossana Sánchez, MD

Title of Presentation

Parental Mosaicism and Monogenic Epilepsies:
Case of *CACNA1A* – Associated Ataxia and
Epileptic Encephalopathy

Learning Objectives:

1. Review the pathogenesis of *CACNA1A* – associated disorders
2. Describe the clinical features of *CACNA1A* – associated disorders
3. Discuss reports of parental mosaicism in monogenic epilepsies

Presenter Financial Relationships: NONE

Company/Role: Children's Hospital of Atlanta,
Genetic Counselor II

Commercial support for RSS/Session: NONE
Financial Relationships of RSS Director,
Planning Committee Members and Others: NONE

Grand Rounds/ Research Seminars

- Grand Rounds

- Every other week
- Mondays 8-9 AM
- Schedule posted on Dept. Website
- Local and invited speakers
- Resident will give one grand rounds each year
- ALL VIRTUAL 2020-2021

- Research Seminar

- Alternates with GR
- Mondays 12-1 PM
- Schedule on Dept. Website
- Local and invited speakers
- Residents should try to go. Discuss with Dr. Gambello.
- ALL VIRTUAL 2020-2021

Self-Directed Learning

- 18 months clinical
- 1.5 months laboratory rotations
- 3 weeks vacation each year
- Leaves 3 months for dedicated self-directed learning
- Involvement in one clinical trial
- Opportunities for other projects/Poster and Manuscript Preparation

Research Opportunities

- Clinical Behavioral and molecular characterization of 3q29 deletion syndrome patients
 - Dr. Jennifer Mulle
- Elucidating inheritance patterns of dystonias
- Next-generation sequencing panels for dystonias
 - Dr. Hyder Jinnah
- Newborn screening performance improvement
 - Arthur Hagar, PhD

Research Projects

- Identifying novel ciliopathy mutations using cilia proteome and EGL databases.
 - Tamara Caspary, PhD
- Mining a longitudinal data set for patterns in outcomes of patients with classic galactosemia.
 - Judy Fridovich-Keil, PhD
- Screen for familial hyperlipidemia in EMR using LDL levels
 - Jaime Vengoechea, MD

Research Projects

- Development of a cell-free DNA-based biomarker for methylation status to be used for disease diagnosis and treatment.
 - Peng Jin, PhD
- Clinical definition and gene discovery for skeletal disorder
 - William Wilcox, MD, PhD
- Exploring methylation abnormalities in tuberous sclerosis complex
 - Michael Gambello, MD, PhD

Genetic Clinical Trials Group

- One of the most capable and experienced clinical trial centers for genetic disorders.
- 30 Sponsored clinical trials
- 10 Registries for genetic diseases
- 3 NIH contracts for newborn screening pilot studies
 - Pompe
 - MPS1, X-Linked ALD
 - SMA
- ALL trainees take part in at least one clinical trial

Examples

- **Biomarin ACHON 111-301**
- **Disease: Achondroplasia**
- **PI: Wilcox**
- **SC: Allison Foley**
- **Actual/Proj Enrollment: 9**

A Phase 3 Randomized, Double-Blind, Placebo-Controlled, Multicenter Study to Evaluate the Efficacy and Safety of BMN 111 in Children with Achondroplasia

Neuronext FX-Learn
PI: Amy Talboy;
SC: Jean Luan McColl
Actual/Proj Enrollment: 7

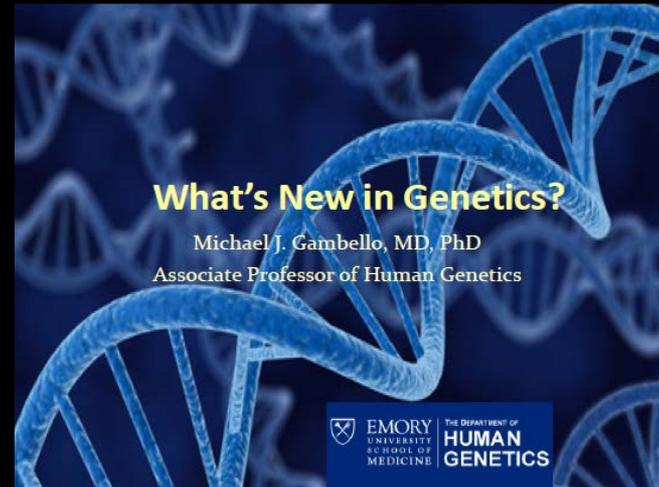
NIH sponsored, Phase 2, Double-blind, Placebo-controlled, Multicenter study for children with fragile X syndrome (FXS) (ages 32 months to 6 years of age - inclusive)
extension

Teaching Opportunities

Pediatric residents
Pediatric neurology residents
Pediatric liver residents
Obstetric-gynecology residents
Medical Students
Genetic counseling students

Educating Primary
Care Providers

Grand Rounds
Invited Lectures
Workshops
Community Outreach
Core Resident Lectures



Meetings

- All trainees go to NAMA first or second year.
- One meeting a year, usually ACMG, SIMD, ASHG.
- Ideal to have an abstract for the meeting.
- Can sometimes get travel award for other meetings e.g. SERGG



Department Retreat

Residents and Fellows are encouraged to attend



Other Aspects of Program

- \$1500 moving expense
- One administrative day/week with no scheduled clinics
- \$600 book allowance
- 3 weeks vacation a year

Email us with QUESTIONS??

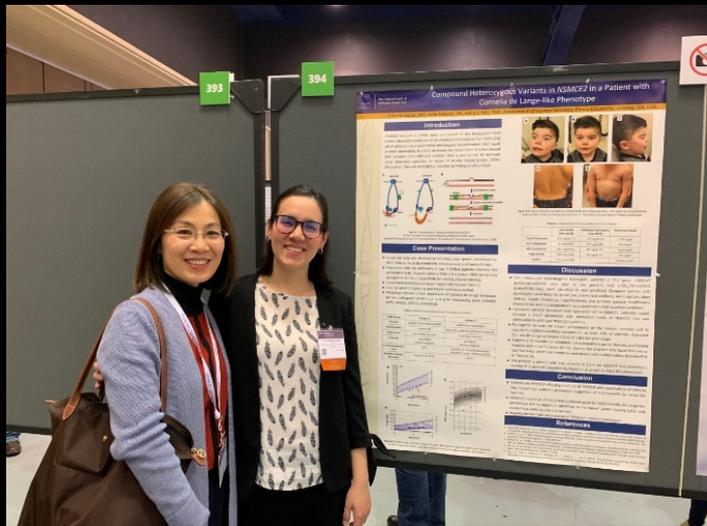
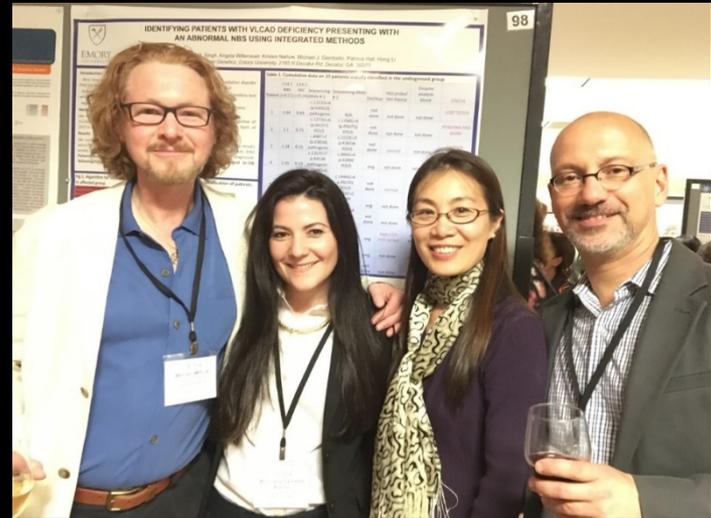


Liz McKenna
Program Coordinator
Elizabeth.mckenna@emory.edu



Michael J. Gambello, MD, PhD
Program Director
Mgambel@emory.edu

We are a Fun Group!



Christmas at Dr. G's

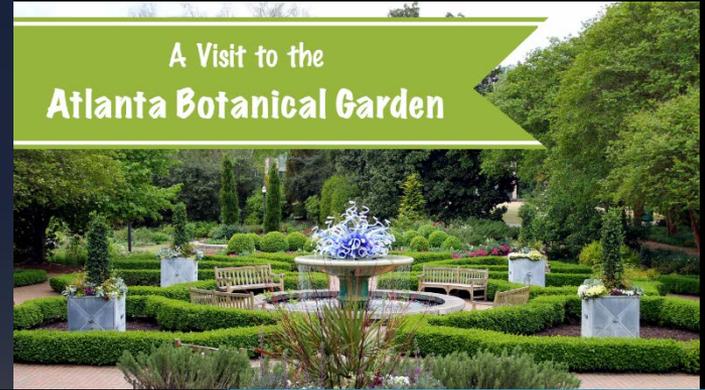


GRADUATION TIME

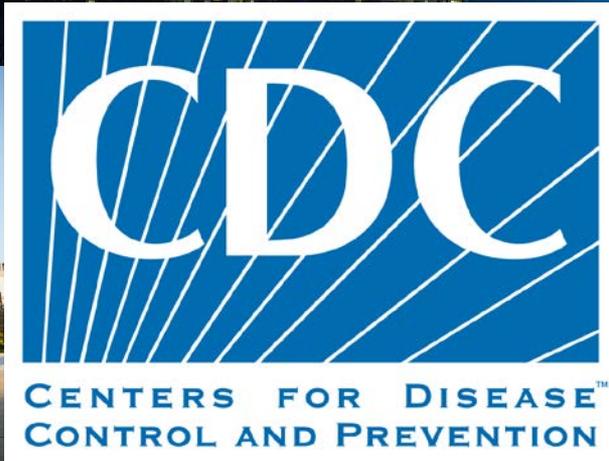


Atlanta is a GREAT City!!!

A Visit to the
Atlanta Botanical Garden



CENTER FOR CIVIL AND HUMAN RIGHTS



CENTERS FOR DISEASE™
CONTROL AND PREVENTION



World's
Biggest
Aquarium

Coca-Cola



DELTA
AIR LINES



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