National Down Syndrome Registry and Biobank: Resources to advance understanding and treatment of Down syndrome

Two recent meetings brought together the Down syndrome community and partners to discuss the need for new resources to help move clinical research ahead quickly and efficiently. These meetings included the Down Syndrome Registry Meeting in September 2010 sponsored by the National Down Syndrome Society, and the Down Syndrome: National Conference on Patient Registries, Research Databases, and Biobanks in December 2010 sponsored by the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD) and the Global Down Syndrome Foundation.

These meetings focused on the need to build new research tools, including a contact registry, a clinical research database, and a biobank. All are resources that will help maximize the efforts of the families who take part in research as well as increase collaborations among scientists, health professionals, and educators.

The purpose of a Down Syndrome Contact Registry is to allow individuals with Down syndrome and their families to register themselves into a database so they can be contacted about clinical research opportunities. This contact registry would include the registrant’s contact information and a short description of their family member with Down syndrome. The contact registry would help researchers identify and recruit individuals who are eligible and might be interested to participate in their clinical research study. This contact registry would also be used to send out information about the progress and findings from each research project. Many times these types of registries are created and maintained by the advocacy community. A structure to govern access to registrants is important. All proposed clinical research projects would be reviewed by experts in the field to determine their potential significance to the Down syndrome community. Registrants would be contacted only after a project is approved.

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The purpose of a Down Syndrome Clinical Research Database is different. Clinical research databases collect, store, catalog, and share information to qualified scientists. They are usually driven by specific research projects that have their own aims. The collected de-identified data (that is, data that does not include names or personal identifiers) can be shared if every project uses standardized collection methods and terms. These clinical databases would provide information about the natural history of Down syndrome throughout an individual's lifespan. This information would help to better understand life transitions and the clinical and medical factors that affect quality of life, medical conditions, and survival. Right now, the data we have are mainly stored in the databases of individual clinicians and investigators. There is no centralized or consolidated clinical research database at this point. The computer technology is simple. The standardization and the resources to push the data into one place are more complicated. But it can be done!

The purpose of a Down Syndrome Biobank is to collect, catalog, store and distribute specific tissues and fluids to approved scientists within the basic and clinical research communities. There are existing biobanks that are specific to a disorder or to a type of tissue (for example, a brain tissue biobank.) There are biobanks that are more general and take any type of tissue from individuals with any disorder. Again, no names or personal identifiers are stored with the sample. Only a minimum amount of clinical data is stored to help understand the source of the tissue. Biobanks are created by private groups (usually non-profit) or by the advocacy communities.

We learned from these meetings that there is a lot of experience in building these resources for other disorders. We do not have to start from scratch. Nonetheless, it will take a large effort from everyone to develop these important resources. Emory and our research partners are committed to building these tools. Representatives from each of our collaborative institutions have attended these meetings and played a large role in the discussions. Our research database is one of the largest in the world. With proper consent, we will share de-identified information through a centralized database with other researchers involved in Down syndrome studies. We also have a large repository of DNA from individuals with Down syndrome and their families. Again with proper consent, we will send samples to the biobank for distribution.

With new scientific and technological advances, scientists are ready to make novel and clinically relevant research discoveries. We think these discoveries will come faster with these centralized resources. In addition to our own work toward this goal, NICHD put out a Request for Information (RFI) to get input from the wider community. One is for the Down syndrome clinical database and registry (NOT-HD-11-002) (if the link doesn’t work, copy the following into the URL bar: http://grants.nih.gov/grants/guide/notice-files/NOT-HD-11-002.html) and another for the Down syndrome biobank (NOT-HD-11-001) (http://grants1.nih.gov/grants/guide/notice-files/NOT-HD-11-001.html). You can go on-line to see this process. Many of you responded with suggestions and support. Everyone’s input is highly valued.

Emory is planning to work with a national biobank to allow other researchers to access biological samples from our current study participants if they consent to be included in the biobank. Please look for a new consent form in the mail soon and contact us if you have any questions about the biobanking opportunity.
The Emory Down Syndrome Project began in 2005 and is currently tracking over 540 families, with approximately 340 complete cases. The purpose of the study is to combine information from interview questions with laboratory data on the behavior of chromosomes to further understand what causes Down syndrome and its related medical problems. More specifically, why do some children with Down syndrome have more medical problems such as heart defects and gastrointestinal defects than others?

Another component of the Emory Down Syndrome Project was the Family Study which included siblings and grandparents of children with Down syndrome. We finished recruitment for the Family Study in 2010 with over 250 families participating. By studying many three-generation families, we will be able to learn more about how chromosomes behave and explore the importance of specific genes on chromosome 21. The biological samples have been sent for genotyping, and the data are being analyzed. Thank you to all the extended family members who made this project possible!

We are also recruiting children with Down syndrome to participate in an NIH-funded study to identify genetic and environmental factors related to congenital heart disease (CHD) in Down syndrome. The goal is to understand why some children with Down syndrome have heart defects and others do not. This study is a collaborative effort between sites at the Kennedy Krieger Institute and Johns Hopkins University, both in Baltimore, MD, Emory University in Atlanta, GA, Oregon Health & Science University in Portland, OR, and Children’s National Medical Center in Washington, D.C. You can read more about their updates on the following pages. Findings from this project will help us to understand congenital heart defects in all children. We have enrolled almost 900 families across sites since 2001.

Our previous work has focused on heart development in individuals with Down syndrome. We have recently started to explore another area of heart research, one related to cardiovascular function. Dr. Cliff Cua is a pediatric cardiologist at Nationwide Children’s Hospital in Columbus, Ohio, who has worked with the Emory Down Syndrome Project for several years. His interest in Down syndrome and research has enabled families, whose child with Down syndrome is treated at Nationwide Children’s Hospital, to learn about and enroll in our studies here at Emory.

Dr. Cua is interested in a particular problem with cardiovascular function – pulmonary hypertension. Pulmonary hypertension is high blood pressure in the lungs. Dr. Cua conducted a study in 2007 in which he found that infants with Down syndrome were more likely than expected to have pulmonary hypertension. Since many infants with Down syndrome have complex medical problems, in particular heart defects, understanding the risk for pulmonary hypertension in the group is important. Working with Dr. Cliff Cua and with Dr. Ken Dooley at the Sibley Heart Center, Children’s Healthcare of Atlanta, we will look for evidence of pulmonary hypertension in infants participating in the Down syndrome studies here at Emory to try to identify risk factors for this condition.
Johns Hopkins University/Kennedy Krieger Institute/Children’s National Medical Center Update

At the Johns Hopkins/Kennedy Krieger Institute sites, we are combining our study recruiting efforts into a single, cohesive project: the Down Syndrome Phenotype Project. Our goal is to reduce the time and effort required for families to participate in all aspects of our research, including the “DS Heart Project,” the “DS Cognition Study,” and “DS Faces.” Combining information from multiple systems (heart, brain, face, GI tract) gives us a better understanding of the mechanisms that underlie the characteristics of Down syndrome.

We’re currently at work setting up social networking sites to make it easier for us to share progress and milestones in the research with the families who made it all possible. In the next issue of the newsletter, look for information about our new Facebook and Twitter sites!!

We’re also planning to be at the International Mosaic Down Syndrome Association (IMDSA) 5th Biennial Research & Awareness Conference at Walt Disney World in Orlando, Florida, (July 8-10, 2011). We’ll have a space reserved, and we’re specifically looking for families with mosaic Down syndrome for a unique study on the function of different types of cardiac cells. If you’re attending the meeting (or just visiting Disney World) stop by and say “hello!”

Enrollment for the DS Heart Project at the combined Johns Hopkins/Kennedy Krieger Institute/Children’s National Medical Center sites has reached a total of 232 families! Thank you to all the families who have been a part of this important work!

Oregon Health & Science University Update

Oregon Health & Science University currently serves as one of five sites for the nationwide Down Syndrome Heart Project. Lead by principal investigator, Cheryl Maslen, Ph.D, we have enrolled over 100 study participants and their families. Most of our recruitment has been conducted through the Down Syndrome Clinic at Doernbecher Children’s Hospital with the help of Joseph Pinter, M.D. Dr. Pinter serves as the Medical Advisor for the Down Syndrome Information Alliance, and his research has focused on neuroimaging in congenital brain anomalies and Down syndrome. We are also currently enrolling children to participate in the national Down Syndrome Cognition Study. Participants in the cognition study are administered a series of computer tests which will provide clinicians with a new way to assess the cognitive abilities of people with Down syndrome. Thanks to all our participants for making this research possible!

If you would like to learn more about the Down syndrome research studies at Oregon Health & Science University, please contact our patient recruitment coordinator, Katy Lesowski at 503-494-7951 or lesowski@ohsu.edu. We will have a booth at the upcoming National Down Syndrome Conference in San Antonio the weekend of August 5th through the 7th, 2011. Please stop by to say hi and learn more about our research!
Emory has joined Pennsylvania State University in a study of phenotypic variation in Down syndrome. Joan Richtsmeier, PhD, and graduate student, John Starbuck of Penn State, continue to collect 3D images of the faces of individuals with Down syndrome and their siblings to explore facial variation caused by trisomy 21. The 3-D images of their typical siblings are being collected as the comparison group. Tracie Rosser, Helen Smith, and Elizabeth Sablon of Emory plan to start enrolling participants in Atlanta later this year. These researchers want to understand how trisomy 21 affects facial development and alters patterns of facial variation within and between different regions of the face (e.g. upper, middle, and lower facial regions.) Preliminary analysis shows that patterns of facial variation are similar for many regions of the face; however, there are differences for the measurement of distances in the lower region of the face and between the middle and lower regions of the face. This suggests that the extra chromosome 21 affects the facial prominences that form, grow, move, and come together to produce the face. This study will begin to help understand facial variation due to trisomy 21 that may lead to problems such as sleep apnea.

Approximately 500 individuals have been enrolled into this study and approximately 500 more are needed. Data collection will continue throughout most of 2011. To participate, a trip to the Image Analysis and Morphometrics Laboratory at Penn State’s University Park campus or to Emory’s Down Syndrome Clinic is required. It typically takes 5-10 minutes per family to collect 3D facial images and complete consent forms. The researchers are willing to consider traveling to locations where groups of individuals may be interested in participating in this study; however, this will depend on how far away the location is and how many people will be enrolled into the study at that location. For more information, please contact Joan jta10@psu.edu or John jms1043@psu.edu at Penn State or Tracie at Emory trosser@emory.edu.

### Going Green

We are planning to send future newsletters by email and have them available on our website in English and in Spanish. Please contact us if we do not have your current email so that we can stay connected. Thanks!! Tracie Rosser 678-422-5284 or trosser@emory.edu.

### Stop in to say Hi!

- **International Mosaic Down Syndrome Association (IMDSA) 5th Biennial Research & Awareness Conference at Walt Disney World, Florida (July 8-10, 2011).**
- **National Down Syndrome Conference in San Antonio (August 5-7, 2011).**
Emory University is part of a nationwide study to understand the differences and similarities in learning abilities among individuals with Down syndrome. This is a continuation and expansion of the pilot study started in 2006. Its purpose is to understand more about how children with Down syndrome learn and problem solve. We are also gathering information about certain medical conditions related to Down syndrome to determine how they may affect learning abilities. Finally, we plan to collect DNA samples to identify genes that play a role in these learning pathways. The other sites participating in the study include: Johns Hopkins University and Kennedy Krieger Institute in Baltimore, MD; University of Arizona in Tucson, AZ; Oregon Heath & Science University in Portland, OR, and The Waisman Center at the University of Wisconsin–Madison, WI. We hope to expand to Children’s National Medical Center in Washington, D.C. later this year. This large scale, multi-site project will have the power to identify factors, both genetic and environmental, that lead to the variation in cognitive functioning seen in individuals with Down syndrome. If we can understand the systems involved in cognition and the factors that play a critical role, we will have a higher chance of developing evidence-based intervention programs. The funding for this project has been provided by the Down Syndrome Research and Treatment Foundation. We are grateful for their support.

Participants come to Emory or one of the other sites for one testing session that may last up to two hours each. Your child will be asked to complete cognitive tests. Most tests will be done on a computer and are just like a computer game. Other tests will be given by a trained researcher. In these tests your child will be asked to sort cards, point to pictures or draw. We will also ask you to complete three short written questionnaires regarding the everyday skills and behavior of your child. We will also need a small blood sample from your child which can be arranged during a routine medically necessary blood draw. We will also need to collect a small saliva sample from both parents if available. We will ask you to sign a form that will give us permission to obtain medical records on your child, to find out if he or she has any of the medical problems often seen in children with Down syndrome.

If you are interested in considering enrolling your child in this study or have questions about the project, please contact:

Tracie Rosser
Phone: 404-778-8474
Email: trosser@emory.edu
2010 was a wonderful year for Down syndrome research! We are proud to be a part of it. Some of the highlights of our projects are listed below, but first we want to thank our participants for all their efforts.

**Cognitive Assessment**—Our cognitive test battery, the Arizona Cognitive Test Battery for Down Syndrome, was published in the Journal of Neurodevelopmental Disorders (Edgin et al., 2010), and highlighted across the web and on NPR [http://kjzz.org/news/arizona/archives/201009/Down_Testing](http://kjzz.org/news/arizona/archives/201009/Down_Testing). We have received funding from the Foundation Jerome Lejeune in Paris to examine the use of our tests in aging adults, and new tests are being designed for use with younger children.

**The Effects of Second Language Learning**—In a study just accepted for publication in the Journal of Intellectual Disabilities Research (Edgin et al., in press), it was shown that moderate exposure to a second language is not detrimental to cognitive development in children with DS.

**Sleep Apnea Effects**—We have just been funded by the Thrasher Medical Research Foundation for three years to complete sleep studies and determine effects of sleep apnea in Down syndrome! We are still recruiting for sleep studies in Arizona. Many families have gotten useful feedback post-adenoid and tonsillectomy surgery or information regarding the presence of apnea.

**Understanding Variability in Outcome**—We are a site of the Down Syndrome Pheno-type Project (DSPP), recruiting individuals 11-18 years for assessments of cognitive function, health and genetic correlations of these outcomes.

**Work on Down syndrome is on the move**! In November we presented four posters at the Society for Neuroscience meeting in San Diego, CA. Over 60 abstracts focusing on DS were presented, with the first-ever symposium with Down syndrome as the sole topic. A round table event sponsored by the Down Syndrome Research and Treatment Foundation also brought researchers together to discuss the next steps. We have research opportunities for individuals with Down syndrome from age seven through adulthood in Arizona. We can sometimes travel to the western states. Call 520-626-0244 or email jedgin@email.arizona.edu.
Maternal age and risk for trisomy 21 assessed by the origin of chromosome nondisjunction: a report from the Atlanta and National Down Syndrome Projects.

Allen EG, Freeman SB, Druschel C, Hobbs CA, O'Leary LA, Romitti PA, Royle MH, Torfs CP, Sherman SL.

Abstract: We examined the association between maternal age and chromosome 21 nondisjunction by origin of the meiotic error. We analyzed data from two population-based, case-control studies: Atlanta Down Syndrome Project (1989-1999) and National Down Syndrome Project (2001-2004). Cases were live born infants with trisomy 21 and controls were infants without trisomy 21 delivered in the same geographical regions. We enrolled 1,215 of 1,881 eligible case families and 1,375 of 2,293 controls. We report four primary findings. First, the significant association between advanced maternal age and chromosome 21 nondisjunction was restricted to meiotic errors in the egg; the association was not observed in sperm or in post-zygotic mitotic errors. Second, advanced maternal age was significantly associated with both meiosis I (MI) and meiosis II (MII). For example, compared to mothers of controls, mothers of infants with trisomy 21 due to MI nondisjunction were 8.5 times more likely to be >or=40 years old than 20-24 years old at the birth of the index case (95% CI=5.6-12.9). Where nondisjunction occurred in MII, mothers were 15.1 times more likely to be >or=40 years (95% CI = 8.4-27.3). Third, the ratio of MI to MII errors differed by maternal age. The ratio was lower among women <19 years of age and those >or=40 years (2.1, 2.3, respectively) and higher in the middle age group (3.6). Lastly, we found no effect of grand-maternal age on the risk for maternal nondisjunction. This study emphasizes the complex association between advanced maternal age and nondisjunction of chromosome 21 during oogenesis.

Investigation of factors associated with paternal nondisjunction of chromosome 21.

Oliver TR, Bhise A, Feingold E, Tinker S, Masse N, Sherman SL.

Abstract: Previous studies on relatively small samples of individuals with trisomy 21 caused by paternally derived errors have shown that: (1) advanced paternal age is not a risk factor for chromosome 21 nondisjunction (NDJ), (2) absence of recombination, but not the location of recombination is associated with paternal NDJ, and (3) there is an excess of males among live-births with paternally derived trisomy 21. An excess of males is also observed among all individuals with trisomy 21. Using 128 families that had a child with trisomy 21 due to a paternally derived error, we examined: paternal age, recombination and the male/female sex ratio. We genotyped STRs along 21q to identify the origin of the error and the location of recombination on the paternal chromosome. Results showed that 32% of paternal meiotic errors occurred in meiosis I (MI) and 68% in meiosis II (MII). We confirmed the lack of a paternal age association with either type of error (mean paternal age for controls, MI, and MII errors: 31.3 +/- 6.6, 32.2 +/- 6.3, 30.6 +/- 6.5, respectively). However, contrary to previous findings, we did not find altered patterns of recombination among paternal MI or MII errors. We found an increased male/female sex ratio among paternal (1.28, 95% CI: 0.68-1.91) and maternal (1.16, 95% CI: 1.02-1.33) meiotic errors. While the sex ratio among individuals with paternal errors was not statistically significant, these findings suggest that selection against female fetuses with trisomy 21 may contribute to the excess of males observed among all individuals with trisomy 21.
Development and validation of the Arizona Cognitive Test Battery for Down syndrome.


Abstract: Neurocognitive assessment in individuals with intellectual disabilities requires a well-validated test battery. To meet this need, the Arizona Cognitive Test Battery (ACTB) has been developed specifically to assess the cognitive phenotype in Down syndrome (DS). The ACTB includes neuropsychological assessments chosen to 1) assess a range of skills, 2) be non-verbal so as to not confound the neuropsychological assessment with language demands, 3) have distributional properties appropriate for research studies to identify genetic modifiers of variation, 4) show sensitivity to within and between sample differences, 5) have specific correlates with brain function, and 6) be applicable to a wide age range and across contexts. The ACTB includes tests of general cognitive ability and prefrontal, hippocampal and cerebellar function. These tasks were drawn from the Cambridge Neuropsychological Testing Automated Battery (CANTAB) and other established paradigms. Alongside the cognitive testing battery we administered benchmark and parent-report assessments of cognition and behavior. Individuals with DS (n=74, ages 7-38 years) and mental age (MA) matched controls (n=50, ages 3-8 years) were tested across 3 sites. A subsample of these groups were used for between-group comparisons, including 55 individuals with DS and 36 mental age matched controls. The ACTB allows for low floor performance levels and participant loss. Floor effects were greater in younger children. Individuals with DS were impaired on a number of ACTB tests in comparison to a MA-matched sample, with some areas of spared ability, particularly on tests requiring extensive motor coordination. Battery measures correlated with parent report of behavior and development. The ACTB provided consistent results across contexts, including home vs. lab visits, cross-site, and among individuals with a wide range of socio-economic backgrounds and differences in ethnicity. The ACTB will be useful in a range of outcome studies, including clinical trials and the identification of important genetic components of cognitive disability.

Variation in folate pathway genes contributes to risk of congenital heart defects among individuals with Down syndrome.


Abstract: Cardiac abnormalities are one of the most common congenital defects observed in individuals with Down syndrome. Considerable research has implicated both folate deficiency and genetic variation in folate pathway genes with birth defects, including both congenital heart defects (CHD) and Down syndrome (DS). Here, we test variation in folate pathway genes for a role in the major DS-associated CHD atrioventricular septal defect (AVSD). In a group of 121 case families (mother, father, and proband with DS and AVSD) and 122 control families (mother, father, and proband with DS and no CHD), tag SNPs were genotyped in and around five folate pathway genes: 5,10-methylenetetrahyrdofolate reductase (MTHFR), methionine synthase (MTR), methionine synthase reductase (MTRR), cystathionine beta-synthase (CBS), and the reduced folate carrier (SLC19A1, RFC1). SLC19A1 was found to be associated with AVSD using a multilocus allele-sharing test. Individual SNP tests also showed nominally significant associations with odds ratios of between 1.34 and 3.78, depending on the SNP and genetic model. Interestingly, all marginally significant SNPs in SLC19A1 are in strong linkage disequilibrium (\( \text{r}^2 > 0.8 \)) with the nonsynonymous coding SNP rs1051266 (c.80A>G), which has previously been associated with nonsyndromic cases of CHD. In addition to SLC19A1, the known functional polymorphism MTHFR c.1298A was over-transmitted to cases with AVSD (P=0.05) and under-transmitted to controls (P=0.02). We conclude, therefore, that disruption of the folate pathway contributes to the incidence of AVSD among individuals with DS.
The Emory Down Syndrome Center, established in 2003, includes education, research, and an important clinical component, the Down Syndrome Clinic. The primary goal of the clinic is to meet the needs of individuals with Down syndrome and their families.

For those of you who are not familiar with our services, we'd like to give you an introduction and answer some of your questions.

What is the age limit?

While our long-range goal is to see individuals of any age who have Down syndrome, we currently have the following limits:

New patients. Birth to age eight years

Returning patients. We have raised our age limit so that children who we first saw before age three years can continue to return to clinic after their third birthday.

How do I make a clinic appointment? To make an appointment, please contact Heather Clark at 404-778-8484 or hmclark@emory.edu. Spanish-speaking families can call Elizabeth Sablon, our medical interpreter, at 404-778-8476. We think it is important that parents are the ones to make the appointment. The initial phone call gives us the opportunity to explain the clinic and determine if there is information about Down syndrome that the family needs immediately. If so, we can often provide it by phone or mail prior to their clinic appointment.

How is a visit to the Down Syndrome Clinic different from a visit to a pediatrician? We are not a substitute for a pediatrician. As with any child, your goal should be to select a pediatrician whom you trust to provide all the best general pediatric care and will be available for those midnight earaches! Our clinic combines genetics and developmental pediatrics.

What does a clinic visit include? A review of your child’s medical history. When parents schedule an appointment, we ask for permission to get their child’s medical records. Information from the birth hospital, pediatrician, and any specialists helps us get to know your child. For example, we can make sure that all recommended tests such as a hearing screen, thyroid test, and cardiac evaluation have been completed adequately.

A discussion of your child’s chromosome report. Parents often want to know more about how Down syndrome occurs, what an extra chromosome 21 means for their child, and if there is an increased chance for another child with Down syndrome in the family.

A physical exam. We complete a basic physical exam of your child and make a special effort to answer any questions you have about features characteristic of Down syndrome.

A developmental evaluation. Our medical director, Dr. Jeannie Visootsak, is a board-certified developmental pediatrician. After conducting a developmental evaluation, she discusses her findings with parents. Recommendations are made for the timing and frequency of early interventional therapy (physical, occupation, speech/language.) Each child is an individual with developmental strengths and challenges. Our goal is to identify these strengths and challenges and make recommendations to maximize each child’s potential.

An explanation of the Healthcare Guidelines for Children with Down Syndrome. These national guidelines provide parents and physicians with a concise outline of special items of care and their timing (e.g., cardiac evaluation, vision and hearing exams, thyroid tests.)

Referrals. Based on each child’s medical history, physical examination, and developmental evaluation, appropriate specialists and/or therapists are suggested if needed.

Answer questions. This is perhaps the most important part of your visit. We urge parents to come with their questions. Each family who visits our clinic is in a different place in terms of their knowledge of Down syndrome and their understanding of what the diagnosis means for their child. We try to tailor visits to each family’s needs.

Where is the Down Syndrome Clinic located?
The clinic is located in our new facility just off the Emory campus near the corner of North Decatur and Clairmont Roads. The address is 2165 North Decatur Rd., Decatur, GA 30033 and parking is easy!

How do I find out more about the clinic? Heather Clark, Clinic Coordinator, will be glad to answer questions related to the clinic, 404-778-8484, hmclark@emory.edu).

All of us in the Down Syndrome Clinic thank the Down Syndrome Association of Atlanta for its continuing support. We couldn’t do it without them!

Jeannie Visootsak, MD
Heather Clark MS, CGC
Adrienne Perkins, M.Ed, MS
Helen Smith, BSN
Elizabeth Sablon, BSW
The Emory Down Syndrome Clinic is excited to introduce two new members of our clinic team. Heather Clark, MS, CGC, is a board-certified genetic counselor and has worked in the Emory University Department of Human Genetics for the past seven years. Heather obtained her Bachelor of Arts degrees from Augustana College in Rock Island, Illinois, where she studied biology, pre-medicine, and sociology. She obtained her Master of Science degree in Medical Genetics from the University of Cincinnati in Ohio.

Heather’s previous clinical work focused on prenatal and reproductive genetics, pediatric genetics, and a group of specialty genetic disorders called lysosomal storage diseases. She is also an experienced study coordinator for numerous clinical trials investigating new treatment medications for lysosomal storage diseases.

Heather is responsible for scheduling families for clinic. As you may know, we ask that families call directly to make their child’s appointment. This gives us an opportunity to get acquainted and explain what is involved in a typical clinic visit. The clinic currently serves children with Down syndrome from birth through age eight years. For more information about the Emory Down Syndrome Clinic or to schedule an appointment, please contact Heather Clark at 404-778-8484 or hmclark@emory.edu.

Adrienne Perkins, M.Ed, MS, is a certified school psychologist and doctoral student in Georgia State University’s doctoral program in School Psychology. She obtained her Bachelor of Science degree in Biology from Bennett College in Greensboro, North Carolina, and her Master of Science degree from the University of Dayton in Ohio.

Adrienne also earned a Master of Education degree in School Psychology at Georgia State University where she will obtain her Education Specialist degree in December. Adrienne has eight years of professional experience as a secondary science teacher. She also has two years of experience conducting psychoeducational evaluations for local school districts.

Adrienne is responsible for performing psychological assessments related to current Down syndrome research projects. She also serves as an educational consultant for families of children with Down syndrome by identifying educational resources and reviewing Individualized Education Plans.

Welcome Heather and Adrienne to the Down Syndrome Clinic!
Both father and mother have 46 chromosomes or 23 pairs, including 2 copies of chromosome 21. Here we show only the pair of chromosome 21.

Occasionally, an egg or sperm is formed with an extra copy of chromosome 21. This example shows the extra chromosome forming in the egg.

Union of this egg with a normal sperm leads to a child with 3 copies of chromosome 21 - trisomy 21 Down syndrome.

Our Mission Statement

Trisomy 21, the leading cause of Down syndrome, occurs when a child receives three copies of chromosome 21 instead of the usual two copies. This is almost always due to a chromosome error during the formation of either the egg or the sperm (see figure at right). Our first goal is to discover how these errors occur and identify factors that affect this process.

Secondly, we want to understand why an extra chromosome 21 causes Down syndrome. We hope to identify specific genes on chromosome 21 that alter development and produce the intellectual disability, congenital heart defects, and other health problems associated with Down syndrome.

Our hope is that increasing knowledge about Down syndrome will benefit families, educators, and health professionals.