Atlanta Down Syndrome Research Conference
A Great Success

By Jeannie Visootsak, MD

Well over 150 parents and professionals attended the conference to hear about the Next Generation of Targeted Treatments in Down Syndrome.

Approximately 150 professionals and parents of individuals with Down syndrome from the Southeast region gathered on March 10, 2012 at Emory University School of Medicine for the Atlanta Down Syndrome Research Conference. The conference was sponsored by the Down Syndrome Association of Atlanta and the Down Syndrome Clinic at Emory University. The attendees were excited about this opportunity to hear from the leading experts the details of ongoing clinical trials and the strides that are being made to assess and possibly improve memory and cognition in individuals with Down syndrome.

We began the day with presentation from Dr. Omar Khwaja, Translational Medicine Leader for Down syndrome clinical trials at Roche. Dr. Khwaja discussed the current Phase I clinical trial which is designed to assess the safety and tolerability of the GABA antagonist drug called RG1662. Parents also learned about the path to drug approval and that clinical trials involving new drugs are classified into preclinical research and four phases. The morning a session on "Overview of Down Syndrome Research" included presentations by Dr. Roger Reeves from Johns Hopkins University School of Medicine, Dr. Stephanie Sherman from Emory University, Dr. George Capone from the Down Syndrome Clinic at Kennedy Krieger Institute, and Blythe Crissman from Duke University. The focus of this session emphasized how studies on trisomic mice models in the laboratory were instrumental in paving the way for human trials to assess some of these potential therapies.

How to reach us

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In Blythe Crissman’s lecture on “Past Challenges, Current Momentum, and Future Avenues,” families were eager to hear about the previous and current clinical trials (e.g., Rivastigmine, Donepezil) in Down syndrome at Duke University.

The afternoon session focused on "Public Policy and Practical Applications" as related to Down syndrome research. Jim Kucik from the Centers for Disease Control and Prevention and Dr. Jamie Edgin from the University of Arizona discussed the policy and applications including the development and validation of cognitive assessment tools for individuals with Down syndrome.

The day ended with a panel of Dr. Jeannie Visootsak from the Emory Down Syndrome Clinic and four parents (Vivian Galletly, Deslie Quinby, Zoila Martinez, and Dan Martin) with children with Down syndrome and who are also professionals serving individuals with Down syndrome as they discussed their thoughts about the topics presented during the day. Although our current Down syndrome clinical trial is in the early stages, parents are nonetheless enthusiastic to learn that the lives of their children may be enhanced with the future psychopharmacology treatment targeting memory, cognitive, and behavior. The speakers emphasized that medications may optimize the individual’s functioning and potential, and allow for integration in the educational and social environment, but medications should always be combined with intervention therapies (e.g., speech therapy), behavioral strategies, and educational support and resources.

This conference is a shining example of the collaboration of parents, clinicians, scientists, scientific experts from pharmaceutical companies, and policy makers joining together to enhance the quality of life for individuals with Down syndrome and their families.

Please do not hesitate to contact us if you have any questions or if you would like additional information about the Roche RG 1662 clinical trial in adults with Down syndrome (age 18-30 years) that is currently being conducted at Emory University. Jean Luan, Down Syndrome Research Coordinator, can be reached at (404) 778-8619.

Welcome Meagan Smith, MS

Please join us in welcoming Meagan Smith, MS, to The Emory Clinic Department of Human Genetics! Meagan will be the new genetic counselor and research coordinator with the Down Syndrome Clinic and Fragile X Syndrome Clinic. Her office will be in Room 145 on the first floor and she can be contacted at 404-778-8528 and meagan.smith@emoryhealthcare.org.
The collaborative Down Syndrome Heart Defects Project (DSHP) recently completed a study that is a step towards understanding why some children with Down syndrome have heart defects and others do not. The research group at Oregon Health & Science University (OHSU) in Portland, OR, led by Dr. Cheryl Maslen, studied genetic differences between children with DS and atrioventricular septal defect (AVSD, sometimes call “hole in the heart”) and children with DS and no heart defect.

Every cell in our body contains instructions for all the necessary development of organs and their function. For example, the cell contains instructions to make an eye, an ear, a heart, even an entire person. These instructions are written in our DNA. DNA is a molecule made up of a small pieces of information organized into genes. Genes function like words within the instructions: used in different combinations they can form sentences and paragraphs. Genes must function properly in specific combinations to form complex living things. Small differences in a gene, known as genetic variation, lead to some of the differences we see among people, such as differences in height. They also lead to an increase or decrease in risk for medical conditions such as birth defects or cancers.

We are specifically interested in genetic variation that may contribute to the chance of a baby with DS being born with a heart defect. Many of the genes involved in the development of the heart are known. We looked at genetic variation in 26 of these genes in over 100 children with DS and an AVSD and over 100 children with DS and no heart defect. We found more rare genetic changes in these genes among children with DS and an AVSD compared to children with DS and no heart defect. Interestingly, the genes with rare variants in the AVSD group are known to work together in a group. This group, or molecular pathway, is called the VEGF-A pathway. This now gives us a good place to start our next set of studies. We will focus on this pathway to find out its role in heart development and how those genetic changes alter that development.

Our results also suggest that, just as every child is unique, the developmental pathway that led to a heart with an AVSD may also be unique. That means we have a lot more work ahead of us. But these results give us the knowledge of what tools we need to get the job done. Recruiting more families into the study will be crucial to future studies.

This work has recently been accepted for publication in the American Journal of Human Genetics:

Acknowledgments:


Helen Smith and Tracie Rosser recruiting for the DS heart study at the Southeast Pediatric Cardiovascular Society meeting in Atlanta.
Dr. Stephanie Sherman and researchers from the Department of Human Genetics at Emory University School of Medicine have been conducting research on Down syndrome for over two decades. Previous studies include the Atlanta Down Syndrome Project (1989 to 1999) and the National Down Syndrome Project (2001 to 2005). Current studies include the Emory Down Syndrome Project and the Congenital Heart Defects Study.

The Emory Down Syndrome Project began in 2005. Recruitment for this study is still in progress and to date nearly 700 families are enrolled in the study with over 400 having completed the study. The purpose of the study is to combine information from mothers’ interviews with laboratory data on the behavior of chromosomes to further understand the causes of Down syndrome and its related medical problems. More specifically, we want to know why some children with Down syndrome have medical problems, such as heart defects and gastrointestinal defects, and others do not.

A component of the Emory Down Syndrome Project was the Family Study which included siblings and grandparents of children with Down syndrome. Recruitment for the Family Study was completed 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. Data from this study are currently are being analyzed.

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 a total of almost 900 families since 2001.

Our work on heart development in individuals with Down syndrome has allowed us to identify other researchers and physicians interested in learning more about Down syndrome. 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.

Research cannot be successful without support from the medical community, intensive effort by researchers, and most importantly commitment of motivated families. We are extremely grateful for the ongoing involvement and willingness of families to take time out of their busy lives to participate in a research study that will someday benefit others. Thank you so much to all who have participated.
Emory University is part of a nationwide study to understand the differences and similarities in learning abilities among individuals with Down syndrome. 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, Children’s National Medical Center in Washington, D.C., and The Waisman Center at the University of Wisconsin–Madison, WI. Children’s Hospital of Philadelphia and the MIND institute in CA.

We have currently enrolled 120 participants across all sites with 38 families completing the study. 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 two testing sessions separated by three months, that may last up to two hours each. Participants 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 participants will be asked to sort cards, point to pictures or draw. We will also ask parents to complete some short written questionnaires regarding the everyday skills and behavior of their child. We will also need a small blood sample from all participants 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 parents to sign a form that will give us permission to obtain medical records on participants, to find out if he or she has any of the medical problems often seen in children with Down syndrome. Several sites are experimenting with road trips to other locations if there are enough families that want to participate. Emory testers have traveled to Alabama, JHU testers went to Virginia, and University of Arizona staff went to California for testing weekends.

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
2012 has been a wonderful year for DS research! We are proud to be a part of it.

**DSA CARES (Down syndrome Arizona- Clinical Care, Advocacy, Research and Education)**

In December of last year we established DSA CARES- a university-community partnership to bring research and resources to the DS community across Arizona. Some early highlights of the center include the addition of supervised training in assessment for graduate students in our lab as well as the inclusion of individuals with DS as summer volunteers in the lab.

**Cognitive Assessment**

Our cognitive test battery, the Arizona Cognitive Test Battery for Down syndrome, has been used in a number of research studies assessing the impact of interventions. We are working on a battery of tests for younger children.

**Sleep Apnea Effects**

Sleep is very important to learning, and children and adults with DS are quite prone to obstructive sleep apnea. We are funded by the Thrasher Medical Research Foundation and DSRTF to complete sleep studies and determine effects of sleep apnea in DS. We are still recruiting for sleep studies in AZ. Many participating families have gotten useful feedback post-adenoid and tonsillectomy surgery or information regarding the presence of apnea. A profile of our work aired on PBS and can be accessed here: https://www.azpm.org/p/top-health/2011/6/27/1830-sleep-and-down-syndrome/

A recent participant commented on our work: “As a little boy, Adam would snore loudly and he always seemed tired and grumpy. His ENT suggested removing his tonsils and adenoids. After the surgery, we noticed some improvement with the snoring and breathing, but he was still tired and irritable. When DSRG asked if we’d like to have our kids participate in a sleep study, we said yes. When we received the results from the DSRG - we were told we should follow up with his pediatrician and have a formal sleep study done. Adam was diagnosed with OSAS and prescribed a CPAP machine. Who would have thought that a little machine like that could make such a big difference! He no longer snores, he's happier, and more alert, and we've been able to cut back on his medication for ADHD. We are very grateful to the DSRG. They have made a big difference in Adam's life, which has made our lives better too”.

**Brain Basis for Cognitive Deficits**

Through the generous funding of the Down syndrome Research and Treatment Foundation and Research Down Syndrome we have initiated studies addressing the brain basis of memory deficits in DS. The results of this work will help us to better understand the source of the behavioral deficits and could serve as biomarkers in clinical trials.

**We look forward to another productive year at this ground-breaking time for DS!**
OHSU is part of two multi-site studies on Down syndrome. First, the Down Syndrome Cognition Project is focused on understanding how children with Down syndrome learn and problem solve. Second, the Down Syndrome Heart Project is focused on identifying genetic and environmental factors related to congenital heart disease in Down syndrome. Dr. Cheryl Maslen serves as the principal investigator of these studies at OHSU while Dr. Joseph Pinter, director of the Down Syndrome Program at OHSU Doernbecher Children’s Hospital, has been an integral part of the recruitment of study participants. To date we have recruited over 100 children with Down syndrome and their parents for these studies. We are currently enrolling children aged 11 to 25 years to participate in the Down Syndrome Cognition Study. Participants will be asked to complete two testing sessions where they 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.

If you would like to learn more about the Down syndrome research studies at OHSU, please contact our recruitment coordinator, Jessica Hunter, at 503-494-1652 or hunter@ohsu.edu.

Thanks to all our participants and their families for making this research possible!
**Genetic modifiers predisposing to congenital heart disease in the sensitized Down syndrome population.**

Li H, Cherry S, Klinedinst D, DeLeon V, Redig J, Reshey B, Chin MT, Sherman SL, Maslen CL, Reeves RH.

**Abstract**

**BACKGROUND:**

About half of people with Down syndrome (DS) exhibit some form of congenital heart disease (CHD); however, trisomy for human chromosome 21 (Hsa21) alone is insufficient to cause CHD, as half of all people with DS have a normal heart, suggesting that genetic modifiers interact with dosage-sensitive gene(s) on Hsa21 to result in CHD. We hypothesize that a threshold exists in both DS and euploid populations for the number of genetic perturbations that can be tolerated before CHD results.

**METHODS AND RESULTS:**

We ascertained a group of individuals with DS and complete atrioventricular septal defect and sequenced 2 candidate genes for CHD: CRELD1, which is associated with atrioventricular septal defect in people with or without DS, and HEY2, whose mouse ortholog (Hey2) produces septal defects when mutated. Several deleterious variants were identified, but the frequency of these potential modifiers was low. We crossed mice with mutant forms of these potential modifiers to the Ts65Dn mouse model of DS. Crossing loss-of-function alleles of either Creld1 or Hey2 onto the trisomic background caused a significant increase in the frequency of CHD, demonstrating an interaction between the modifiers and trisomic genes. We showed further that, although each of these mutant modifiers is benign by itself, they interact to affect heart development when inherited together.

**CONCLUSIONS:**

Using mouse models of Down syndrome and of genes associated with congenital heart disease, we demonstrate a biological basis for an interaction that supports a threshold hypothesis for additive effects of genetic modifiers in the sensitized trisomic population.

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**Altered patterns of multiple recombinant events are associated with nondisjunction of chromosome 21.**


**Abstract**

We have previously examined characteristics of maternal chromosomes 21 that exhibited a single recombination on 21q and proposed that certain recombination configurations are risk factors for either meiosis I (MI) or meiosis II (MII) nondisjunction. The primary goal of this analysis was to examine characteristics of maternal chromosomes 21 that exhibited multiple recombinant events on 21q to determine whether additional risk factors or mechanisms are suggested. In order to identify the origin (maternal or paternal) and stage (MI or MII) of the meiotic errors, as well as placement of recombination, we genotyped over 1,500 SNPs on 21q. Our analyses included 785 maternal MI errors, 87 of which exhibited two recombinations on 21q, and 283 maternal MII errors, 81 of which exhibited two recombinations on 21q. Among MI cases, the average location of the distal recombination was proximal to that of normally segregating chromosomes 21 (35.28 vs. 38.86 Mb), a different pattern than that seen for single events and one that suggests an association with genomic features. For MII errors, the most proximal recombination was closer to the centromere than that on normally segregating chromosomes 21 and this proximity was associated with increasing maternal age. This pattern is same as that seen among MII errors that exhibit only one recombination. These findings are important as they help us better understand mechanisms that may underlie both age-related and nonage-related meiotic chromosome mal-segregation.
Our Latest Publications

Neurodevelopmental outcomes in children with Down syndrome and congenital heart defects.

Visootsak J, Mahle WT, Kirshbom PM, Huddleston L, Caron-Besch M, Ransom A, Sherman SL.

Abstract

Trisomy 21, the chromosomal condition responsible for Down syndrome (DS, OMIM #190685), is the most common identifiable genetic cause of intellectual disability. Approximately half of all children with DS are born with a significant congenital heart defect (CHD), the most common of which is an atrioventricular septal defect (AVSD). As children with comorbid DS and CHD increasingly survive cardiac surgery, characterization of their early developmental trajectories is critical for designing early interventions to maximize individual potential. Herein, the developmental domains (cognitive, language, and motor) of children with DS and AVSD (DS + AVSD, n = 12) were compared to children with DS and a structurally normal heart (DS - CHD, n = 17) using the Bayley Scales of Infant and Toddler Development III. The DS + AVSD cohort mean age was relatively the same as controls with DS - CHD, 14.5 ± 7.3 months compared with 14.1 ± 8.4 months, respectively. Although the motor domain was the only domain that showed a statistically significant difference between groups (P < 0.05), both cognitive standard scores (P = 0.63) and language composite standard scores (P = 0.10) were lower in the DS + AVSD cases compared with the DS - CHD controls although it is not statistically significant. Since this is the first study to examine the early developmental outcomes of children with DS + AVSD, the findings may be useful for clinicians in providing anticipatory guidance.

See Complete article in:

SOURCE: Department of Human Genetics, Emory University, Atlanta, Georgia, USA. Jvisoot@emory.edu

Down syndrome: national conference on patient registries, research databases, and biobanks.


Abstract

A December 2010 meeting, "Down Syndrome: National Conference on Patient Registries, Research Databases, and Biobanks," was jointly sponsored by the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD) at the National Institutes of Health (NIH) in Bethesda, MD, and the Global Down Syndrome Foundation (GDSF)/Linda Crnic Institute for Down Syndrome based in Denver, CO. Approximately 70 attendees and organizers from various advocacy groups, federal agencies (Centers for Disease Control and Prevention, and various NIH Institutes, Centers, and Offices), members of industry, clinicians, and researchers from various academic institutions were greeted by Drs. Yvonne Maddox, Deputy Director of NICHD, and Edward McCabe, Executive Director of the Linda Crnic Institute for Down Syndrome. They charged the participants to focus on the separate issues of contact registries, research databases, and biobanks through both podium presentations and breakout session discussions. Among the breakout groups for each of the major sessions, participants were asked to generate responses to questions posed by the organizers concerning these three research resources as they related to Down syndrome and then to report back to the group at large with a summary of their discussions. This report represents a synthesis of the discussions and suggested approaches formulated by the group as a whole.

See Complete article in:

SOURCE: Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health, Bethesda,
Lack of maternal folic acid supplementation is associated with heart defects in Down syndrome: a report from the National Down Syndrome Project.

Bean LJ, Allen EG, Tinker SW, Hollis ND, Locke AE, Druschel C, Hobbs CA,

Abstract

BACKGROUND:
Maternal folic acid supplementation has been associated with a reduced risk for neural tube defects and may be associated with a reduced risk for congenital heart defects and other birth defects. Individuals with Down syndrome are at high risk for congenital heart defects and have been shown to have abnormal folate metabolism.

METHODS:
As part of the population-based case-control National Down Syndrome Project, 1011 mothers of infants with Down syndrome reported their use of supplements containing folic acid. These data were used to determine whether a lack of periconceptional maternal folic acid supplementation is associated with congenital heart defects in Down syndrome. We used logistic regression to test the relationship between maternal folic acid supplementation and the frequency of specific heart defects correcting for maternal race or ethnicity, proband sex, maternal use of alcohol and cigarettes, and maternal age at conception.

RESULTS:
Lack of maternal folic acid supplementation was more frequent among infants with Down syndrome and atrioventricular septal defects (odds ratio [OR], 1.69; 95% confidence interval [CI], 1.08-2.63; \( p = 0.011 \)) or atrial septal defects (OR, 1.69; 95% CI, 1.11-2.58; \( p = 0.007 \)) than among infants with Down syndrome and no heart defect. Preliminary evidence suggests that the patterns of association differ by race or ethnicity and sex of the proband. There was no statistically significant association with ventricular septal defects (OR, 1.26; 95% CI, 0.85-1.87; \( p = 0.124 \)).

CONCLUSIONS:
Our results suggest that lack of maternal folic acid supplementation is associated with septal defects in infants with Down syndrome. Birth Defects Research (Part A), 2011. © 2011 Wiley-Liss, Inc.
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 the 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 totailor visits to each family’s needs.

Where is the Down Syndrome Clinic located?

The clinic is located on our new facility just off the Emory campus near the corner of North Decatur and Clairmont Roads. The address is 2165 North Decatur Road, 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
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Return Service Requested

Si desea copia en español, por favor llame a la Sra. Elizabeth Sablon al 404-778-8476.

**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.

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The father's sperm have 1 copy of each pair of chromosomes, including chromosome 21.

The mother's eggs have 1 copy of each pair of chromosomes, including 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.