

THE EARLI Edition

The Newsletter of the EARLI Study

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Spotlight on the Pontiflet Family:

Life is an Adventure

Four years ago, 2-year-old Tiarra Pontiflet was diagnosed with Autism Spectrum Disorder (ASD). Her diagnosis provided her parents, Breanna and Rodney Pontiflet Jr., with an answer to their escalating concerns about their daughter's development and behavior. Since Tiarra's diagnosis, Breanna and Rodney have become very familiar with autism research. In addition to joining a prior study, they are currently participating in the EARLI Study at the UC Davis MIND Institute (Medical Investigation of Neurodevelopmental Disorders) in Sacramento, Calif.

Breanna recalls she and her husband first became concerned about Tiarra's development when their daughter stopped reaching developmental milestones. She did not walk or talk when expected, and when she finally did begin to speak, she only repeated others' words. Realizing these behaviors were not typical, Breanna and Rodney Jr. turned to their pediatrician for advice. They were referred to the Regional Center at California's Department of Developmental Services, which provides assistance for individuals with developmental disabilities. It was there that Tiarra officially received her ASD diagnosis. Since her diagnosis, the Regional Center has provided Tiarra with intensive speech and occupational therapy. With this assistance she has been able to flourish. Breanna reports Tiarra's memory is her strong suit: "It's incredible!"

After receiving Tiarra's ASD diagnosis, the Pontiflet family was referred to the MIND Institute. There, they immediately enrolled their second daughter, Tatyanna, in the Infant Sibling Study, which also focuses on early ASD detection. When Tatyanna was 18 months old, Breanna and Rodney Jr. received the surprising news that they were expecting again! To Breanna's astonishment and Rodney Jr.'s excitement, the ultrasound revealed not one heartbeat, but two!

When Breanna was six weeks pregnant with the twins, she received a call from the EARLI Study asking if the family would like to participate. Breanna and Rodney Jr. didn't have to think twice. They were strongly motivated to join the study because of their experience raising their oldest daughter with ASD. As Breanna explains, "I want to be supportive and help as much as I can. ... We want to better understand ASD, find the cause of it, and support the research of it in any way we can."

EARLI has monitored the Pontiflets' twins, Rodney III and Tavia, since their birth. Their infectious smiles light up the clinic at every visit. When Rodney III came in for a 21-month courtesy clinic visit, he was diagnosed with ASD. Tavia also received a diagnosis a few months later. The family reports Rodney III and Tavia's diagnoses have not drastically changed their daily lives. Although Tiarra, Rodney III and Tavia's behavior differ in many ways, Rodney Jr. feels that already having a child on the spectrum has helped them know what to expect.

Through the EARLI Study, the Pontiflet family is helping researchers to pinpoint factors that may contribute to ASD. However, the study also provides the family with peace of mind of knowing specialists are closely observing their children's development. As Rodney Jr. reiterates, "early detection is key," and they know EARLI ensures that their children will have the best prospect for treatment and growth. Both parents describe their lives as an adventure. They see their children's diagnoses less as a disability and more as an opportunity. With this in mind, Breanna put it most eloquently: "We are hectic and busy, but a lot of fun."



Rodney Jr. with all 4 kids. From left to right Tatyanna, Rodney III, Tavia, and Tiarra. Mom's behind the camera trying to help get all the kids' attention.



INVESTIGATOR'S VOICE

As the director of the EARLI site at Kaiser Permanente in Northern California, I am very excited to be involved in this groundbreaking research study. My interest in studying autism stems from both my professional and personal experiences.



Lisa with her nephew Eytan.

On the professional side, I am a perinatal epidemiologist, and for my whole career I have had an interest in understanding how factors related to pregnancy and delivery impact the growth and development of children. On the personal side, I have a nephew with autism, who was diagnosed in the late 1980s at age three. At that time, there was very little research on autism outside of the realm of genetics. By the late 1990s, I decided to shift my research focus from the study of congenital birth defects to studying autism full-time.

Over the past 15 years, there has been an explosion of research into autism from every field of science. We have learned a great deal more about how genetic factors influence development, and we have a much greater appreciation for the important role that environmental factors play. We now recognize that autism is not a single condition, but rather there are many 'autisms,' and each may have a unique set of causes and risk factors.

Why is the EARLI Study so important? The EARLI study was designed to take an in-depth look at the many different genetic and environmental factors that may be related to the development of autism spectrum disorders all at the same time. By collecting such comprehensive information and biological samples in 'real-time' – during pregnancy and the first few years of life – we have an accurate account of what happened during these critical periods of development. With this information, we will be able to identify specific sets of factors that are related to specific developmental trajectories for children with autism. This knowledge is critical not only for understanding the causes of the many 'autisms' but for developing effective treatments.

I feel very fortunate to be able to contribute to the science and improve our understanding of Autism Spectrum Disorders. Thank you for participating in this important study and giving so generously of your time. Together, we have a real opportunity to find answers and help families all around the world.

Lisa Croen, PhD

Would you like your family featured in an upcoming EARLI Edition? Let your site coordinator know!

FLYING THE FRIENDLY SKIES

Trapped by reality, FREED by imagination.

By Michelle Landrum

Clarkie was amping up, his agitated whine growing louder, his head swaddled in his yellow blanket and his hands clamped over his ears. A broken vent hissed out a powerful jet of air above Seat 7E, where my six-year-old struggled to stay in control after a long day of travel, airports and delays.

The malfunctioning air vent -- a simple annoyance for most people -- threatened to push Clarkie into a full-fledged meltdown even before the plane left the gate in Detroit.

Travel tests the patience of any parent, but when your child has an autism spectrum disorder, as the younger of my two sons does, it helps to be resourceful, daring and lucky. Delayed nearly three hours by a mechanical problem, the darkened plane was full of irritable passengers trying to fall asleep. And yet Clark's high-pitched whine began to amplify.

"I can't stand it!" he cried, as the vent hissed unrelentingly.

I called over the flight attendant and explained. "My child has autism, and the noise from this broken vent is unbearable to him. Can it be fixed or can we change seats?"

"I'm sorry, the flight is full," the attendant said, before hustling off to her duties.

As Clarkie's whining continued, tears of frustration filled my older son's eyes, his patience frayed by his brother's all-too-frequent agitation. Desperate, I jammed the yellow blanket against the vent -- silence! --

then considered whether I could hold my arm in that position for sixty-three minutes until we touched down in Baltimore. Then... a glimmer of inspiration.

Fumbling in my purse with my free hand, I found tissues and stuffed them into the vent crevice. A woman across the aisle offered me two Band-Aids to tape the tissues in place. I gratefully accepted, and the mood around us seemed to lighten. Seeing the makeshift repair, a man joked that I should've been called in to fix the plane in the first place. What could have been an emotionally turbulent flight turned into something positive. The rest of the flight was smooth. And later, as Clarkie darted about at baggage claim, passengers recognized him and smiled at his antics.

The two bandage strips covering crumpled tissues made a comical but effective fix for the broken vent -- one that required a measure of ingenuity, desperation and goodwill, much like the life that we manage to pull together every day. For all things about autism that chafe against our family, we travel on, coping with what we can and delighting when we find empathetic people who lend a hand, even when they may not totally understand our difficulties.

Michelle Landrum is one of several EARLI Study staffers who have a personal connection to autism



Adam Bayer (left) is the best friend of his younger brother, Clark, who is on the autism spectrum. Travel can be particularly stressful for Clark -- and the rest of his family.

I cannot always make the world a quiet, easy place for my "Quirky Clarkie" -- nor can any parent do so for a child -- but on this one day, in this one way, I was his hero.

Once the offending vent was silenced, Clark calmly removed his hands from his ears, looked up at the wadded tissues and grinned. "Mommy, you saved the day!"

This essay and dozens more are featured in *Chicken Soup for the Soul: Raising Kids on the Autism Spectrum*.

Dr. Rebecca Landa, an EARLI Co-Investigator in Maryland, wrote the book's forward.



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Privacy Statement

The EARLI Study is grateful to all our families, and we respect your privacy and confidentiality. Any families featured in this newsletter have given their explicit permission to EARLI.

EARLI



Dear EARLI families,

This is a very exciting time for the EARLI study. All of the babies from our 230-plus families are now more than a year old and about one-third have 'graduated' from the study at age three. Now the EARLI investigators can begin to connect the dots between the assessments of your children we have done in our clinics, the many questions you answered in your diaries, and biological samples you provided throughout your participation.

EARLI investigators now have a number of analyses under way and many more in the planning stages. While we don't have any findings from these analyses to share with you yet, I wanted to give you a brief preview of just a few of them here. When the results are in, we will update you with any findings. Now is also a good time to remind you that because the EARLI study is so unique in the type, timing, and intensity of the data we collect, our investigators have the opportunity to ask novel questions and to do analyses that have never been done before. So, when we do have findings from this work, other researchers using data from other studies will have to replicate what we see, perhaps several times, using different study designs and samples, before they confirm anything we first report. This process can take time and requires patience but it is still extremely gratifying to be moving on to this stage with EARLI.

So, what have we begun working on? One set of studies involves the new field of "epigenetics." We all know genes pass on the body's complex instructions from parents to child. You are probably also aware that scientists have shown that a large and very complex group of genes can influence the risk of autism. While instructions in our genes are passed down from parent to child through the genetic code (the sequence of the A, T, C, and G amino acids), scientists now realize the code does not tell the full story of how genes influence the body, including the developing brain. The way our genes are packed and unpacked in our cells, as well as the presence or absence of small molecules that can adhere to our genes, also influences the expression of these genes. In other words, for some genes, two people with the exact same genetic code can have different instructions issued by the genes because of these other "epigenetic" factors. Furthermore, while the genetic code is passed from parent to child according to Mendel's laws, these epigenetic factors are influenced by the environment. So EARLI has begun to look at these epigenetic factors – whether they are correlated with the behavior we are seeing in our EARLI babies and whether they are affected by factors ranging from differences in diet during pregnancy to chemicals in the environment. Research on epigenetics could turn out to be one very important way scientists better understand how environmental factors affect neurodevelopment.

We've also started to look at other factors that may affect brain development. Autism has long been known to be three to four times more common in boys than in girls. But the reasons for this are still a mystery. One theory is that the hormones that differentiate boys from girls could somehow be related to autism risk. Of course, we are not talking about the hormone burst we see in teenagers, because at that age autism will have already long been a part of an affected teenager's life. The hormonal influence on autism risk is most plausible if these hormones affect the brain at its earliest stages of development. But how do we measure hormones that can reach a developing baby's brain before it is born? Believe it or not EARLI is now looking at meconium (that first poop you collected from your baby's diaper) to see if we can measure sex hormones in those samples! Your baby started making meconium at around the thirteenth week of gestation and the hormones we measure in the meconium might reflect the hormones that could actually have reached your baby's brain while you were carrying him or her. If we can measure hormones in meconium, we can then look at factors in the environment that influence hormone levels and also see how hormone levels during prenatal development might influence babies' behavior later in life.

This was a very, very quick peek at just two lines of investigation now underway. Other analyses are also progressing and at this moment EARLI investigators have four different grant applications under review at the National Institutes of Health in the hopes of securing additional funding to start a wide range of other new analyses. None of this would have been possible without you and your family. So, speaking on behalf of all my EARLI colleagues, we continue to be so grateful that you have let us get to know you and your family over these years. Your time, energy, and commitment are now being transformed into truly novel science that we believe will one day give us new insight into autism's causes. We promise to keep you posted as our research moves along at this exciting new stage.

Sincerely,

Craig J. Newschaffer, PhD
Principal Investigator, EARLI Network
Director, A.J. Drexel Autism Institute

"We're depending on the EARLI study to shed some light on the causes of autism. I'm grateful to the participants of the study."

Laura D. Bono
Board of Directors, SafeMinds



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Get to know *EARLI's Graduating Class*

Think back to your favorite year in school... Remember your teacher? Your classmates? The big stuff that happened that year?

You may not have thought of it this way, but you're part of another class now: The EARLI Study cohort. We wanted to give you a sense of who else is in this very special class and how extraordinary you are for sticking with EARLI for almost four years!

First off, you're a select group: 233 mothers and 175 biological fathers. You all have a great deal in common – welcoming a new baby while caring for an older child on the autism spectrum. Many of you are caring for more than two children at home.

Perhaps unsurprisingly, most of your older children who have an Autism Spectrum Disorder (ASD) are boys (198 male vs. 35 female), which matches the sex disparity in ASD diagnosis seen generally in the United States. This gap is one of the big mysteries in autism research, and the data you have provided to EARLI will help shed light on this mystery.

More than half of you – 120 families – live in California, where EARLI has two research sites.

The rest of you enrolled through EARLI's other two research sites on the East Coast: 45 of you currently live in Maryland, 40 in Pennsylvania, 17 in New Jersey, 5 in northern Virginia, 2 in Washington, D.C., 2 in Delaware, and 1 in Connecticut. We even have 1 family overseas!

We're happy that the EARLI families reflect the racial and socioeconomic diversity of the United States. You span the spectrum in terms of income and educational backgrounds. The EARLI mothers, most of whom are in their late 30s, are quite a diverse group: Almost 60% report their race as white, while 10% are Asian, and almost 12% are black. Many report being multi-racial. Almost 17% report having Hispanic or Latina ethnicity. Some of you are single mothers, and others have blended families.

You've stuck with EARLI through major life changes. Many families have moved households during their time in EARLI – including some families who moved to other states or overseas but continue to participate.

The 240 babies born into the EARLI study really aren't so little anymore: a dozen of the children are now older than age 4, and the youngest baby has already turned 18 months old. Their



genders are almost evenly split between boys (128) and girls (112).

A handful of EARLI families welcomed twins, and two families enrolled in EARLI for consecutive pregnancies.

We offer you our sincere admiration and thanks for taking part in EARLI. You all deserve applause for helping to advance the understanding of autism. When it comes to this graduating class, you make EARLI a success!