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re-search

Tracking Rare Incidence Syndromes Project: Helping Parents Advocate for Children with Rare Medical Condition



Over the last ten years, Dr. Deborah Bruns, Associate Professor in the Department of Educational Psychology and Special Education, has published in a wide range of professional and medical journals regarding children with rare trisomy conditions such as trisomy 18 and trisomy

9 mosaic. Dr. Bruns is also an expert in the area of feeding issues for children with disabilities, having recently co-authored a book on the subject. She is active in state and national organizations advocating for children with disabilities and their families, and is the Principal Investigator for the Tracking Rare Incidence Syndromes (TRIS) project, which has collected longitudinal health, medical, developmental, family support and demographic information from over 500 families from around the world.

Her career began in New York City, where she took a job as a classroom teacher working with children with multiple disabilities. Among these children were several who had been diagnosed with trisomy 18. Trisomy conditions are a genetic anomaly in which a gene pair, the blueprint for how the body develops, includes a redundant copy. The most common form of the disease is Down's syndrome, where this occurs on chromosome 21. However, the condition can involve other chromosomes. Dr. Bruns research has centered on the "orphaned" conditions, particularly trisomy 18, 13, and 9 mosaic.

"It sometimes surprises people to find out that we've come to this field through special education, rather than genetics or some medical field. But special ed is where a lot of the difficulties come to light regarding how these conditions interact with everything else



Infant with trisomy 18 exhibiting a clenched hand, often characteristic of the condition.

that a person is: their emotional self, their cognitive self, their physical self," Dr. Bruns recently reported. Her background as an educator has occasionally created difficulties. "Sometimes at conferences I am at a disadvantage because I'm not from that medical background, and people may question whether I should be playing with the 'big boys.'" However, her focus on the children has allowed her to connect with many families that include a member with one of these conditions, which, in turn, has led to a steady stream of introductions to additional participants.

"My sensibilities are more at the family level. Most parents, if they take out a picture of their child, get a response along the lines of 'She's so cute' or 'He's adorable.' The parents I work with frequently get silence and stares because people don't have a frame of reference for how to respond."

One of the more important aspects of the project is educational, not just of parents with affected children, but also of medical personnel who may never have come across a patient with one of these conditions before, or who are not aware of the range of outcomes parents have experienced. "There's a mother, who's an anesthesiologist in Norway whose child with trisomy 18 lived for just three days. In Norway, they usually don't want to treat a child like that at all. They're very hands off. But, this time, they messed with the wrong mom. Now, even though her daughter is not with us anymore, she's working with a group of us, internationally, to change the perspective of the medical profession so that they see these children as children, and not just as diagnoses." The mother has taken her daughter's information and data from the TRIS project and is disseminating it through the medical community there. "Our [TRIS project] literature is out there. Parents take it with them to meetings with their doctors."

Medical professionals and new parents are



Proud parents hold their infant child, who is receiving oxygen.



The gift of touch.



Parents bonding with their child through physical contact.

becoming aware of Dr. Bruns and the project. “We’re being called on to consult in cases, and even when we don’t get the result we would want for a family, the network of parents keeps expanding as parents we’ve worked with put other parents that they meet in touch with us. Sometimes, parents come up against an authority that wants to deny some treatment because they say it won’t work such as cardiac surgery, but we have evidence of cases where it has.”

The TRIS project has been collecting data on children in these circumstances for seven years. Most published studies on children with these diagnoses have been case studies based on a single patient that a doctor has seen. Often, they have only followed the patients until they went home from the hospital. Since the patient may not follow up with that same doctor or clinic, these authors may not know the ultimate outcome. There hasn’t been this kind of longitudinal data available that the TRIS project collects. We contact parents annually for updates on their living child regarding changes in medical status, educational and therapy services, family changes and developmental progress.

Currently, there are three people working on the project, Dr. Bruns, her undergraduate research assistant, Emily and Shirley, the mother of an adult with a rare form of trisomy living in California (partial trisomy 6p). Shirley takes care of following-up with project participants through the project’s website and other electronic means.

“From our data, we’ve found amazing things, like two families living in the same hamlet in the Netherlands. In a community with, maybe, 200 residents, these families have children with the same condition and who are just two years apart in age, yet they didn’t know each other. We’ve found several pockets of children with these diagnoses in the United

States, as well. There’s one in Texas and another one up in the Chicago area.”

Funding for TRIS has come from family support organizations and individual donations, because the larger organizations, such as the NIH, tend to favor projects coming from the medical community. “We keep going with small donations here and there.”

One obstacle this causes relates to travel. “In order to go to the conferences each year, I have to fund that travel through the project. Many of the conferences are medically oriented. Since I don’t quite fit their profile, even though we’re collecting data that we know they need, I don’t always have the option of formally presenting our findings, which limits the avenues I have for funding my attendance.”

Because the project has become the hub of a community of families, it has created opportunities to encourage people touched by Trisomy conditions to tell their story. “I’ve been in touch with the mother of one of our children who is a medical ethicist, specializing in palliative care. I’m hoping we can get her to write up her case because she can speak with authority from both the parental and the medical perspective.”

Sometimes, Dr. Bruns will be recommended to a pregnant woman whose child has been diagnosed in utero. This can create a delicate balancing act. “I have to let them know that their child might not make it, while at the same time giving them ammunition to go in with if their doctor doesn’t know that trisomy cases are not by definition hopeless. Working from hospital or insurance standards, a doctor may not want to treat a child with trisomy 13, particularly if they are also presenting certain other conditions.” This can leave a pregnant woman, who is dealing with the stress of what the trisomy condition will mean for her child and her family, having to prepare to advocate that child’s

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position with a doctor who may not believe help is possible. "At the same time, I have to prepare her for the possibility that the worst case scenario may come to pass. I try to make sure that she has the evidence she needs in case a doctor pushes back. I also encourage her to find out what her OB will do in different situations, to learn what her insurance will cover, and to prepare her for the fact that the child she will have will not be the child she envisioned before receiving the diagnosis."

The project tries to find answers when parents call to find out whether a particular medical issue is related to their child's trisomy diagnosis. Recently, when a mother was being charged in court with neglect, the project was able to pull data that showed that specific dental conditions are common in children with trisomy 13 and did not indicate neglect. As of now, there is no centralized listing of what dental conditions are commonly seen in these cases, but Dr. Bruns and her assistant are writing their findings up for a specialized dentistry journal.

It is not unusual for a parent's involvement with the community to continue even after the death of their child. A mother in St. Louis found Dr. Bruns shortly after her son's death. They arranged a meeting where she talked at length about her son and asked Dr. Bruns about survivors. She became very involved in the community, and was on the committee for a recent conference in St. Louis. She and Dr. Bruns have spoken at several presentations highlighting her son and her experiences with his care. Dr. Bruns has assisted her in writing a book about her experiences, which was published in 2012.

People ask Dr. Bruns about life expectancy, but, at this point, she doesn't feel she can answer those questions. "Articles and other literature suggest that

there is a less than a 10% likelihood of surviving the first year in these scenarios, but we seem to be pulling in a lot of those 10%, if that's the case." Nothing in the data gives a definitive answer of why this should be. It may be the treatment they receive, or the lack of certain complicating factors "There's a brain anomaly issue and a heart issue that we know have an effect in the trisomy 13 cases. But with the 18's, it's less clear. Some have a heart problem that has been known to resolve on its own without surgery. I do not know why my sample would be so biased, but I have more kids in the program who are alive than I do that have passed."

The TRIS project has completed surveys on over 300 children and adults with rare trisomy conditions that lived more than two months (with over 70% still alive at survey completion) and approximately 170 surveys completed for infants who have passed (stillborn or within 60 days of birth). "One of the girls in the study had her Sweet 16 last year, and will be 17 soon. We've got a whole bunch of teenage girls right now. It is difficult to know how representative the project's sample is. Parents who are grieving," Dr. Bruns notes, "who lost their child before they heard about the project, are unlikely to fill out a survey." Despite that issue, the project has a great deal of data on a relatively large sample of children. "We have 75 children and adults who have full trisomy 18, as opposed to mosaic or partial forms which are less severe, so just running that demographic data . . . well, if I could afford three more assistants we could be churning research out like crazy. Instead, right now, we have to pick and choose what studies we have the resources to complete. Comparable studies typically focus on hospital registries or country-wide data collection on live births and deaths for these groups."

Until the day that funding becomes available, Dr. Bruns and her assistant will keep trying to answer parents' questions when they call. 3

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If you would like additional information on the Tracking Rare Incidence Syndrome project, please visit the project's website at: <http://web.coehs.siu.edu/grants/tris>.

An Interview

Learning The Ropes: An Undergraduate's Introduction To The Craft Of Research

by Alan Overton



Emily Campbell is a fifth year senior in the COEHS studying Special Education. She is also the research assistant to Associate Prof. Deborah Bruns in the Department of Educational Psychology and Special Education. I recently had the chance to sit down with her to discuss what it was like to work so closely with a professor on a research project while still an undergraduate student.

How did you start working with Dr. Bruns' and her Trisomy project?

EMILY Dr. Bruns came in to one of my introductory special education classes and gave us a five-minute spiel, saying that she was looking for a research assistant. She said that if any of us were interested in the position and wanted to put in an application, we should come by her office.

What made you want to tackle this kind of research?

EMILY When she talked about it, I didn't really understand all it would entail, but I thought it sounded interesting, so I decided to give it a shot. I credit taking this position with actually spurring my interest in research.

Are there any particular skills that you have had to develop to use in research beyond what you use as a student?

Honestly, I was never much of a writer before, but, since I started working on the TRIS project (SEE PREVIOUS STORY) I've seen my writing skills improve significantly, and have had professors in my other classes comment to that effect. Dr. Bruns has also asked me to maintain the project's spreadsheets, so I've been called on to do some data mining and create graphics for presentations.

Have you had to add any Math courses to handle data analysis?

EMILY So far, because of the type of data we've been collecting and the kind of questions we've been asked, most of the Math we've had

to do has been along the lines of sorting counting, and calculating simple averages on our data.

Do you see yourself continuing in research as a career?

EMILY Definitely. Last year I was a part of the undergraduate symposium that's held here on campus every year in April. This year I'm on the board of the committee that's putting the symposium together for students here at SIU. Last year, I was talking with my father about how I had made a poster and done a presentation, and he sort of dared me to take it to the next level. He texted me and said, "I can't wait for Dr. Emily Campbell to come home." My immediate reaction was that it sounded ludicrous, but, after thinking about it, I decided, "I want to do that!"

Do you think you will continue to do research into disability and special education?

EMILY I'm not sure. I love the research that we're doing now. I love the families and the people we're involved with, and my brain is in that mode now where I wake up thinking, "I could do this, or I could do this," but I don't really know where I want to go. It may take some more searching to find that, I think.

My major is Special Education, and I think that once I get out into the world, I'll be better able to see what my strengths and interests are, and see where I might best be able to help expand on what we know. Since I'll graduate in December, I would probably be limited to substitute teaching for the

first few months after I finish this degree. I'll have some time to get my bearings. One thing that's always interested me was the Peace Corps, so I need to decide whether I would want to do that right out of school or, whether I should try to find a teaching position. My timeline for returning to school will probably depend on what opportunities present themselves.

Was research something you expected to study when you first came to college?

EMILY When I first got to college, I didn't understand how many new things to which I would be exposed. I started out at the Culinary Institute. When I switched to the College, here, it was to prepare to be a high school Chemistry teacher. Later, as I found out more, I considered Special Ed. But it feels that everything that I've done has gone into building the me who I am now.

So, what specifically are you working on this semester?

EMILY We have a list of case studies to document, and I'll be doing one each month through the end of the year. We're writing up the information that we presented at a conference last fall and writing an article that's just looking at the developmental data we've collected on Trisomy 13 and 18, plus we've got a couple of other irons in the fire that are not really full ideas yet. We've got a conference coming up in July where we'll be presenting a compilation of the small studies we've done throughout

over the last two semesters. We're in touch with a lot of families who will call us up and say something like, "My kid has asthma. How many other kids with Trisomy 16 have asthma?" They ask just so that they can figure out whether or not the asthma is related to their trisomy diagnosis. We've collected a lot of data like that, so we can try to compile it for them to answer specific questions like that. So we're pulling those ad hoc reports together to present to a wider audience.

Do you have any advice for undergraduates who might be considering getting involved in research at this level?

EMILY Well, this is my favorite job of the ones I've had, but you really have to have the mindset that you want to help other people by progressing a whole field of inquiry. You can't be scared of exploring uncharted waters. You can't be afraid to make mistakes. Sometimes I'll write up something for Dr. Bruns and I'll get it back covered in red ink. That doesn't even faze me anymore. You have to be resilient and willing to learn from what other people say about your writing. ☺

Dr. Bruns joined the discussion, and I asked her to comment on whether there were any differences between graduate and undergraduate researchers.

Dr. Bruns Students, particularly undergrads are open to new ideas. I can throw something at them and say, "Give it a shot," and they'll dive in. They'll try anything. My assistants have all been willing to be mentored and supervised, but they still have that motivation and initiative that comes from the novelty of the whole situation. The best student researchers I've had have been confident enough to give me a poke to get something done if it's holding up the next step for them. For me this is good because I'm always juggling multiple projects and can lose track of what I need to do, even if I'm carrying the article I need to work on around with me.

On the TRIS project, we work with many of our parents over Facebook, and Emily has never had any issue with jumping online with a parent. She understands the project and she knows when to ask for help, so if she gets back with a parent first about a question, that's fine. If someone needs data that she can put together, she does it. She's even leading one of our family groups. I see great things in her future. ☺