



Resource Article

Inside this edition

“Supporting Families
Through the Diagnosis Process -
Looking Closer at Rare Conditions”

Resource Article	1
What do the data say?	3
Consultation Corner	4
On the WWW	6
Continuing Education	6

As the work to build the human genome continues, genetic testing is becoming increasingly important. What information can genetic testing provide? How do genes influence a child’s typical or atypical development? When should genetic testing be considered and when should it not be an option? How can genetic testing help us understand a child’s condition? These types of questions come up more and more as children experience lengthy developmental plateaus, demonstrate notable skill regression, or engage in seemingly unusual behaviors. Yet, many neurologists and psychiatrists are reluctant to order genetic testing. Salm, et al. (2014) examined this subject and presented their results in the article, *Use of genetic tests among neurologists and psychiatrists: Knowledge, attitudes, behaviors, and needs for training.*

The researchers sampled 535 neurologists (163) and psychiatrists (372) who responded to an email invitation to be a part of the study. The original invite went out to the American Medical Association master list of professionals who

opted-in to receive surveys. This master list included 2,167 neurologists and 5,316 psychiatrists and nearly 10% participated in this study. The neurologists were asked 52 questions and psychiatrists were asked 62; psychiatrists were asked additional questions about the genetic basis for several psychiatric disorders. Survey questions included varied response options, such as dichotomous (Yes/No, Agree/Disagree), multiple choice, Likert scaled responses (e.g., How would you rate your knowledge of genetics 1 – Very Poor to 5 – Very Good), and some questions allowed for further elaborated responses. The survey took approximately 15-20 minutes to complete.

Analysis of the survey responses supported the need for additional training and familiarity with genetic testing as well as greater networking surrounding the topic of genetics. A notably greater number of neurologists than psychiatrists ordered genetic testing in the last six months, 74% and 14% respectively. Patients broached the topic of direct-to-

Resource Article (continued)

consumer genetic testing with 15% of study respondents. Respondents reported ordering genetic testing for the following considerations:

- Neuromuscular disorders (33.4%), such as Muscular Dystrophy
- Movement disorders (32.1%), for movement disorders such as Huntington's disease
- Dementia (6.6%), such as Alzheimer's disease
- Stroke (6.2%)
- Pediatric neurological conditions (5.7%)
- Pharmacogenomics, for the purpose of understanding how genes affect a patient's response to medications (2.1%), such as in the condition of epilepsy

Most respondents in this study felt that greater use of genetic testing was needed (68% of those who had used genetic testing and 55% of those who had not). Only 33% of respondents felt confident about how and where to order genetic testing. Nearly half of the neurologists (49%) and three-quarters (75%) of the psychiatrists reported they do not have a genetic counselor to whom they would refer their patients. Interestingly, about half of all respondents felt that genetic testing could result in psychological harm; no difference was found between those who had ordered genetic testing and those who had not.

Neurologists and psychiatrists, and patients for that matter, are showing an increasing interest in genetic testing and a growing curiosity for

learning how it can be used diagnostically and in treatment. Yet, the knowledge gap about how genetic testing can help, when it should be conducted, and where to refer to ensure access to genetic testing continues. Additional coursework related to genetics is being included in medical school curricula. Additionally, online modules and continuing medical education opportunities at conferences are helping to provide additional avenues for learning and advancing learning about the who, what, when, where, why, and how regarding genetic testing.

Even though most children below the age of three are not seeing psychiatrists, there are some children who see neurologists when developmental questions arise. In these cases, and as a consumer of medical services, it is in our best interest to consider our physician's experience with and ability to order genetic testing and counseling.

Salm, M., Abbate, K., Appelbaum, P. Ottman, R., Chung, W., Marder, K., Leu, C-S., Alcalay, R., Goldman, J., Curtis, A., Leech, C. & Taber, K. (2014). Use of genetic tests among neurologists and psychiatrists: Knowledge, attitudes, behaviors, and needs for training. *Journal of Genetic Counseling*, 23(2), 156-163.



What do the data say?

What are reactions to infant death?

Infant mortality has been associated with the greatest and longest lasting stress in parents (Christ, Bonanno, Milkinson, & Rubin, 2003). According to the Center for Disease Control and Prevention, about 1/2 of child deaths occur during infancy. Unintended injuries are the leading cause of death in children ages 1 - 14 years. On the upside, the death of children (ages 1-14 years old) now accounts for less than 5% of the deaths in the US. Yet, we know for children with rare and severe conditions infant death can be a difficult reality.

Families who lose a child due to complications with a rare or complicated diagnosis have to integrate their loss into the future fabric of their lives. Christ et al. (2003) referred to infant death as “the death of the parent’s future dreams” (Intro., para 2). Bereavement, grief, and mourning encapsulate this very sad experience. Bereavement encompasses all the experiences associated with death including the anticipation, the death, and all the subsequent actions and adjustments that follow. It is most certainly a complex and dynamic experience loaded with different emotions and reactions. Grief is part of this and is highly individual. It can be expressed in many ways and can complicate one’s ability to do everyday tasks. Further complicating the process is the internal process of mourning that is also expressed through varied actions and behaviors.

Losing a loved one, especially a very young child, is a highly confounded process that brings out various emotions and reactions. As EI providers supporting families of very young children, it is helpful to understand common reactions to the loss of a child while also recognizing that bereavement is incredibly individual and circumstance specific.

According to Christ and colleagues a common reaction to the passing of an infant is an “intense preoccupation with thoughts and images of the dead baby” (Christ, et al., 2003, Grief Reactions, para 1). Other reported reactions include having a continued sense of the baby still being alive, hearing the baby cry, or feeling of the baby kicking as he might have during the pregnancy. Still other reactions include feeling uncertain about the

safety of themselves and their family, feeling angry, irritable, scattered, and short-tempered. It is common too for parents to extend blame to others, such as providers, health care professionals, and even spouses, while searching for answers. Moreover, when the search for answers comes up empty, the frustration grows leading to further disappointment. These emotions can be internalized and result in depression, decreased self-esteem, and guilt, which can last for years ebbing and flowing as time passes and memories slip in and out.

Interestingly and important for EI providers, “many parents of infants who die report being particularly stressed by people who avoid any discussion of the loss or offer clichés of dismissing statements such as ‘you can have another child’” (Christ et al., 2003, Grief para 2). While it is impossible to define the best response, it seems that being responsive and following the family’s lead regarding further correspondence is a good start.

Yet, this too is complicated as there can be differences with parent’s grief reactions. Mothers were found to have longer lasting and more intense reactions to the loss of an infant while fathers experienced greater denial immediately following the baby’s passing (Wing, 2001). There is also a possible impact on the parent’s relationship. Feelings of conflict, anger, and blaming are common for both parents. Yet, incongruent expression of these emotions can lead to communication breakdowns, including avoiding discussions about the baby and minimizing the provision of emotional support and shared coping. Marital conflict in the mix of grieving the loss of the baby is a difficult reality that parents might have to work through as they learn to live without their precious baby. Understanding these possible dynamics and being aware of community supports are essential for EI providers, especially when a family they know or work with has lost a loved one.

Christ, G. H., Bonanno, G., Malkinson, R., & Rubin, S. (2003). Bereavement experiences after the death of a child. In M. J. Field & R. E. Behrman (Eds.), *When Children Die* (Appendix E). Washington, DC: The National Academies Press. Accessed from: <https://www.nap.edu/read/10390/chapter/18>



Consultation Corner

From September through December 2016 we are excited to have **Dr. Deborah A. Bruns** as our Consultation Corner expert. This series will explore *Supporting Families Through the Diagnosis Process and Looking Closer at Rare Conditions.*

I've spent the last few months writing about how early intervention providers can assist parents with a child with a rare diagnosis. I've reiterated the importance of information, resources, and support. I wanted to use this final article to share the voice of parents to add another layer, a more in-depth view, on the previous topics. I hope the following parent stories help further our understanding about the journeys parents take when their child has special needs, needs that are complex and affect every area of their lives, as well as the lives of those around them.

Kari's Story

Leila was diagnosed as having trisomy 18 at 22 weeks gestation. We were offered termination, but that was not an option for us. The OB did allow the pregnancy to continue, but wouldn't do anything heroic at birth. We were told that she would be still born. After changing doctors and hospitals, Leila was born by emergency C-section at 34 weeks 6 days and was immediately intubated and taken from the delivery room.

Having a child with trisomy 18 definitely changes the perspective and day-to-day operations of a family. Many outsiders view having a child with special needs as a limiting factor in the spontaneity and freedom allowed to the family. In one sense, this is quite accurate. Having a child with complex medical issues does not allow us to do certain activities, however plenty of new opportunities have arisen.

Having a daughter with trisomy 18 has presented us with several speed bumps, particularly the loss of relationships with certain family and friends who do not support the decisions that we have made regarding Leila's care. At first, these losses were challenging, but they shed a new light on the importance of having meaningful relationships in our lives. *It is more important to have a handful of friends and family who would*

do anything to help you in a time of need, than to have an address book full of individuals who "would love to help you out but can't this time." Our "new normal" also includes spending time getting to know people; giving others the most precious treasure we can give, our attention.

A final component to our "new normal," which is the most significant, is the lessons our children have learned as a result of having a little sister who has trisomy 18. We have always strived to instill character traits in our children. However there has not been a more effective approach than having a little girl who cannot speak but can teach them what it means to be caring, generous, humble, optimistic, persistent, fearless, determined, purposeful, and compassionate.

Our "normal" has definitely changed in the nearly five years since Leila was born. As we reflect back on the changes that have occurred during these five years, both in our family and our society as a whole, we are thankful for this "new normal." Being given the opportunity to show others that all people deserve the chance at life has been perhaps the most powerful experience of our entire lives. The smiles and giggles that Leila shares with everyone she interacts with have been multiplied tenfold onto the faces and attitudes of those who interact with her.

Consultation Corner (continued)

Lynel's Story

It started at 19 weeks when the sonogram went "south" after not locating the head and then as they searched vaginally via sonogram and found the encephalocele. As the doctor whirled into the room with his sad face and placed his hand on my knee, even before he said anything, we knew that we were going to be facing something that would change our lives in an instant. The doctor said, "there is something wrong with the baby; the head did not close in the back. There is a large encephalocele growing behind the baby's head. We are surprised the fetus is still alive at this point."

"She's a FLUKE," this is how I learned we were having a little girl. This was also the one and only explanation the doctor gave me over the phone after viewing the results from an amniocentesis showing that all of Jeff's and my chromosomes were normal. There was that word again, *normal*, but everything was NOT *normal*.

I found people who I surrounded myself with to help, pray, extend friendship, and support me emotionally. I didn't need friends who felt sorry for me, but rather, I needed those who made me laugh and feel as though life was back to *normal* even though, things were definitely not.

One day during a church service as I prayed and felt this baby girl moving, dancing it seemed, joyfully, and it dawned on me to name her Haley Faith. We were definitely having her out of faith, another new *normal*. But, also entering this last trimester, Jeff and I both planned out a funeral service for her, rather than a nursery.

As Haley survived birth and all that surrounded that day, so many prayers were answered for her to hold our pinkies, to nurse, to open her eyes, make sounds, cry. That's all I had asked for, really. But, as she survived and the team from the hospital came to take her for further care, I had to say goodbye after having her for only five hours, and all through that time everyone including the medical staff just

waited. Waited to see if she would pass. What a strange feeling.

We were finally given the news that she would never see, hear, walk, talk, etc., and she'd have very low functioning. She was dismissed from the hospital after only four days in the NICU and we were told to "enjoy her, she'll be gone in two weeks." Those two weeks were tough. Our new *normal* was now different. She was alive, but not a *normal* baby.

I lived in probably more fear at that point. Here we had her, alive and in person, and as each day passed I was getting more attached, and that fear just swept over me of losing her, my new *normal*. But, she was thriving. She was sleeping, eating, pooping, cooing, everything a baby should do. How was she going to die?

Two weeks post-op, to remove the encephalocele, we brought her back to the neurosurgeon. They never ushered us into a clinic room to remove her 40 stitches in the back of her head. Instead they ushered us to a conference type room. There, I could hear the neurosurgeon explain to the 1st year resident doctor that "there is no need for me to see her, she shouldn't be here much longer and I have other patients to tend to. You take her stitches out here and send her home."

My new *normal* was changing again. I realized I needed to speak up for my child. No one was going to do it for me. It's hard to name what one specific new *normal* reaction would be to one diagnosis or condition. We've had to adjust all throughout her lifetime as to what a new *normal* would look like.

So many surgeries, doctors in three states. Neurosurgeons would scratch their heads in bewilderment saying "she's not typical, she's not a *normal* child with this type of defect." That was a new *normal* and remains that way even now. This is because she hears, she walks with a walker, she talks (non-stop), she sees even though technically she does NOT have a visual cortex of the brain, and she functions fairly high. And she just turned 13 in July 2016.



On the WWW

Do you want to learn more about genetic testing? The National Human Genome Research Institute provides a user-friendly informative overview about genetic testing. The resource is online at: <https://www.genome.gov/19516567/faq-about-genetic-testing/>

The site includes answers to the questions about what genetic testing is, what can be learned from genetic testing, what types of testing exists, what are the benefits and drawbacks, and more. Also included are links to other helpful resources

related to genetic testing including a glossary of genetic terms.

You can also tab to a section on family history to create your own family health history. This related resources is part of a 2004 U.S. Surgeon General initiative reinforcing the importance of family health history. The link to My Family Health Portrait is online at:

<https://familyhistory.hhs.gov/FHH/html/index.html>



Continuing Education for KIT Readers

The Comprehensive System of Personnel Development (CSPD) is offering a continuing education opportunity for KIT readers.

In line with the focus on *Supporting Families Through the Diagnosis Process—Looking Closer at Rare Conditions*, readers are invited to receive continuing education contact hours for reading the monthly KIT publications (September - December 2016) and completing a multiple-choice exam about the content covered in these KITs.

KIT readers will receive the exam in January 2017. There is no need to register for the CEUs. Rather, if you are interested complete the exam online at www.edis.army.mil

Upon successful completion of the exam, you will receive a certificate of non-discipline specific continuing education contact hours.

KIT Newsletters
are available
online at
www.edis.army.mil

Thank you for your continued interest in the KIT.

