Educational & Developmental Intervention Services (EDIS) Personnel Development

KIT

Keeping In Touch

SEPTEMBER 2016

Inside this edition

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

Resource Article

When a young child fails to meet milestones, the questions begin. What, exactly, are the developmental milestones for my child's age? Maybe I'm being overly anxious about my child? But when the child's developmental progress stalls over time, the questions increase as does the urgency. Whom should I ask about this concern? Where do I get more information? What's causing the problem? Will she grow out of it? How long before I see changes? Will this affect the rest of her life? What can I do to help? These questions are typically put to physicians, but the answers may be slow to come or result in more testing, questioning and waiting. This is often the case for families with children who have rare diseases and disorders. Paturel (2012) recounts the difficulties families face as they address the uncertainties associated with the diagnostic process of pinpointing a child's disease in her article, 'Too Rare for Research? People with rare diseases often experience significant delays in diagnosis and access to few, if any, treatment options'.

Paturel tells the history of a little girl named, Maia, who appeared typically developing. Yet, when she was unable to sit independently at eight months the concerns came and she was ultimately diagnosed with cerebral palsy (CP). Bloodwork analysis, however, suggested something different. The diagnosis of CP was removed and the family was told Maia had an unidentifiable muscle disease. The family sought out specialists to help them rule out and determine the cause(s) of her developmental failings. Meanwhile Maia started to lose strength leaving her unable to walk or lift her head in the bed. After many appointments Maia was given the diagnosis of alpha dystroglycan – related to dystrophy – a rare form of congenital muscular dystrophy, which is caused by a genetic mutation. Maia was eight years old by the time she finally got her diagnosis.

This example illustrates the challenges faced by families of children with rare disorders. It's not just the families that are at a loss. "As physicians, we're good at identifying common diseases. But when a disease affects fewer than 200,000 people, as rare diseases do, many physicians will never see it" (p.

Resource Article (continued)

30), reported David Robertson, M.D. professor of neurology and pharmacology at Vanderbilt University. Identifying the appropriate diagnosis helps families better understand what is happening with the child and helps with their expectations. It helps providers as they work with families on treatment options and intervention. Additionally, identifying rare diseases helps to contribute to the data base of information on rare diseases.

Because so few people are affected by such diseases, the payoff for developing treatments has been low. Historically, funding for research on rare diseases has been difficult to procure. This is changing as laws have been enacted to secure funding and speed up the time from development to implementation. The Orphan Drug Act, 1983, "…offers incentives to companies who develop new drugs to treat a rare disease" (p. 31) is one such law. In this case, incentives include tax credits and opportunities for grant funding. Unfortunately, however, because of the lack of treatment options for rare diseases, families can be drawn to sources who offer treatments without clinically proven effectiveness and/or FDA approval in the hopes of finding a 'miracle cure'. Some of these experimental treatments (drugs, procedures, and devices) can been ineffective at best and dangerous at worst.

Once the diagnosis has been identified, finding treatment can be challenging. While it's true that large metropolitan areas often have more specialists and research facilities, they don't always have information pertinent to rare diseases. For example, Paturel describes the search for treatment of a family with a child with hypothalamic hamartoma (a rare neurologic disorder, which involves seizures, developmental delays, rage behaviors, and endocrine problems). The family was told that there was nothing to be done and that their son would be likely reside in a group home by the time he was five years. The family moved to Phoenix, Arizona to be close to a facility that specialized in neurological diseases. They found other families with the same disease and through this communication learned about a surgical treatment being performed by a specialist in Sydney, Australia. While the doctors in Phoenix were unwilling to try the procedure because of the risks, the family consulted with the specialist in Sydney, flew to Sydney and had the procedure. It was successful. Their son became seizure-free. And the specialist flew out to Phoenix to perform the procedure on other children suffering the same disease. This was made possible by the persistent networking of one family.

We've often heard that the "squeaky wheel gets the grease". This is certainly apropos for families with children with rare diseases. Without their tenacity, energy and courage, they would have few answers and little hope. As early interventionists, working with families of children with disabilities it is important for us to support their dreams too.

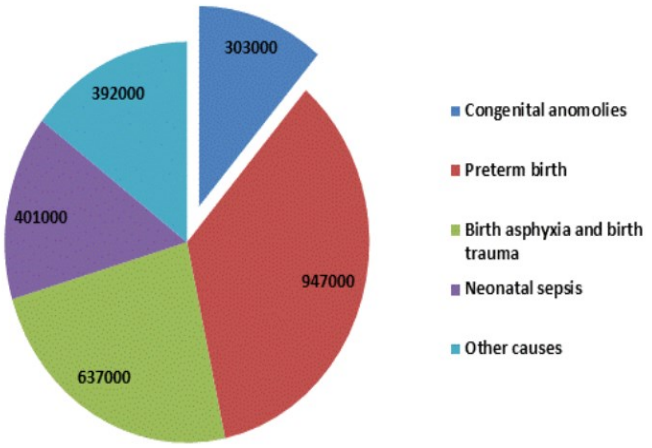
Paturel, A. (2012). Too Rare for Research? People with rare diseases often experience significant delays in diagnosis and access to few, if any, treatment options. Neurology Now, 8 (2), 29-33.

What do the data say?

What is the frequency of congenital anomalies?

Congenital anomalies, known also as birth defects and congenital disorders, include malformations, deformations and chromosomal abnormalities that exist at or before birth. According to the World Health Organization (WHO) congenital anomolies are a common condition, yet there are no estimates defining the number of children born with a serious congenital disorder. Down syndrome is included in the most common and serious conditions covered by the ICD-10 definition of congenital anomalies.

Congenital anomalies can result is long-term disabilities or death. Looking back to 2004, the WHO estimated that about 7% of all neonatal deaths, worldwide, were caused by congenital anomalies (WHO – Sixty-third World Health Assembly, 2010). More recently, WHO estimated that over 300,000 newborns die within 4 weeks of birth, worldwide, due to congenital anomalies (WHO, 2016). Examining child causes of death further the WHO identified the following mortality estimates during the neonatal period in 2015, worldwide (WHO, 2016).



The actual cause of these congenital conditions may be due to genetic connections, or possible infectious, nutritional, or environmental factors, making it difficult to pinpoint actual causes (WHO, 2016). Interestingly, yet likely not surprising, socioeconomic status and demographic circumstances are also factors associated with congenital anomalies. According WHO, 94% of severe congenital anomalies occur in low- and middle-income countries. Compromised access to prenatal care and attention, as well as maternal infections and poor nutrition are attributes that may contribute to this high percentage. Exposure to environmental conditions, such as medication, alcohol, tobacco, and radiation are also associated with congenital anomalies.

Focused on defining interventions to prevent congenital anomalies, the WHO is working many partners, including the US Center for Disease Control and Prevention's (CDC) National Center on Birth Defects and Developmental Disabilities, to improve women's and children's health to help newborns survive and thrive. Early screening, detection, and treatment are also essential intervention considerations. Also among important interventions is the provision of early intervention. By effectively supporting families of children with congenital anomalies, we can help families help their children achieve their greatest potential while supporting as they navigate their journey of having a child with a congenital disorder.

World Health Organization (2016). Congenital anomalies Fact Sheet. World Health Organization. Accessed from: http://www.who.int/mediacentre/factsheets/fs370/en/

World Health Organization (2010). World Health Organization Sixty-third World Health Assembly, Provisional agenda item 11.7, A63/10. Accessed from: http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf?ua=1&ua=1

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.

Deborah A. Bruns is a Professor in the Department of Counseling, Quantitative Methods and Special Education at Southern Illinois University Carbondale. She teaches undergraduate and graduate level coursework in Early Childhood Special Education (ECSE) including assessment and curriculum methods. Dr. Bruns also teaches courses focusing on collaboration with families and related service providers. She has been Coordinator of the Special Education program (undergraduate and graduate levels) since 2010.

Prior to this position, Dr. Bruns worked as an Educational Therapist at the New York Foundling Hospital (Manhattan, NY). She provided services to infants, toddlers and preschoolers with multiple disabilities and special health and medical needs. Several of the young children she worked with there were diagnosed trisomy 18. These experiences led her to launch the Tracking Rare Incidence Syndromes (TRIS) project in 2007. The project is to raise awareness of and improve services and supports provided to children with conditions including trisomy 18, trisomy 13 and trisomy 9 and their families. Data is collected via survey. Results have produced approximately a dozen articles, 20+ presentations and case studies (available for download at http://tris.siu.edu/case-studies/index.html).

Dr. Bruns has also co-authored a book on feeding challenges (available from Brookes Publishing at http://products.brookespublishing.com/Feeding-Challenges-in-Young-Children-P246.aspx). Topics include typical and atypical feeding development, screening and assessment of feeding needs, general feeding strategies and specialized feeding interventions. Dr. Bruns has also presented at the regional, state and national levels on this topic. In addition, she participates in a number of committees and special interest groups within the Division for Early Childhood (DEC) on the topics of leadership and advocacy in ECSE.

Determining Early Intervention Eligibility: Assisting Parents to Navigate the Medical Diagnostic Process

The diagnostic process can be an overwhelming and emotional experience for parents. There is the “need to know” along with the uncertainty and worry in determining something is not quite right with their infant or toddler. A parent or caregiver may be the first to notice a delay in development, atypical behavior and/or a combination of the two. Early Intervention (EI) providers are in a position to assist parents to move forward with taking action about their concerns. They are also uniquely suited to provide support in answering questions, addressing concerns, and, most importantly, helping families navigate the medical diagnostic process. Moving into the realm of medical conditions can be particularly daunting for parents due to unfamiliar jargon, testing and the like.

Consultation Corner (continued)

The Individuals with Disabilities Education Act (IDEA) outlines the EI eligibility process. In addition, there are some differences across states. For example, in some sates, EI operates within the Department of Education, while, in others, the lead agency is the one focused on health and human services. There is also some variation in percentage delay for EI eligibility (e.g., 30% in one developmental area). (The Early Childhood Technical Assistance (ECTA) Center provides state-by-state eligibility information; link is below). In addition, included in the law are conditions which are thought to place an infant or toddler at risk for delays. Here, too, there is some variation in determining which conditions have a high probability of resulting in delayed development and are 'automatically' eligible for EI. Biological risk is one area including infants born before 28 weeks gestation.

A range of disability conditions such as Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and cerebral palsy can immediately qualify a child for EI. For these families, the diagnostic process might have included prenatal diagnosis or testing results at or shortly after birth. Other times, EI providers may be helping families navigate the initial medical diagnostic process. The remainder of this article focuses on helping families navigate this process.

A medical concern is often identified by a medical professional and may evoke fear, confusion and a myriad of other emotions as parents process what may be a long term change to their young child's life (e.g., medication to control seizures, dietary restrictions). It is important for EI providers to be accepting of each individual parent's acceptance or hesitance to accept that their child's diagnosis. Sharing experiences of parents in the same situation can provide support but not all parents seek this type of information during the medical diagnostic process. Some prefer to hear these stories after a definitive diagnosis is made.

In addition, EI providers cannot make a medical diagnosis but they can assist parents in a number of ways. They can help parents articulate their concerns by reinforcing and guiding their efforts to share authentic and specific information with medical professionals. Documentation, verbal descriptions, and video illustrations can be especially important to present concerns or puzzling behaviors such as trouble feeding or posturing during diaper changes and dressing. EI providers can also share appropriate and reliable resources about the suspected or confirmed medical condition(s) (e.g., WebMD, support organization information pages). They can help parents to "connect the dots" when reviewing medical reports and evaluation results as well.

Beyond information sharing EI providers can also help parents identify lingering questions and concerns that will be important for them to share with medical providers during upcoming appointments. An additional and significant support EI providers share during the medical diagnostic process is emotional support. By actively listening and paraphrasing, EI providers can help parents articulate and understand and/or reconcile the feeling they are experiencing.

Consultation Corner (continued)

Throughout the medical diagnostic process, parents should also be repeatedly made aware they are the most central part of their child's evaluation. This can be especially reassuring as parents face a diagnosis that can, potentially, impact their child, family members and caregivers over the long-term. In addition to the presenting medical concerns, parents have knowledge of their infant or toddler's daily routines, favorite activities, responses to unfamiliar situations, etc. This information can assist professionals especially if/when the child is uncooperative during the process. Finally, encouraging parents reinforces the importance of parent participation especially during what is often a stressful time.

Realizing and, eventually, accepting that one's child has a medical condition can have a profound impact on parents and other family members. There is often denial, grief and acceptance (order can vary or one remains as the others are completed but may resurface at a future time). There can be feelings of anger, despair, and, for some parents, relief. Taken together, EI providers are in a unique position to offer guidance and support as parents begin and navigate the diagnostic journey and support their infant or toddlers continued growth and development.

Resources:

http://www.militaryonesource.mil/efmp?content_id=266631

http://ectacenter.org/~pdfs/topics/earlyid/partc_elig_table.pdf

http://www.parentcenterhub.org/repository/journey/

http://www.parentcenterhub.org/wp-content/uploads/repo_items/notalone.pdf

http://www.parentcenterhub.org/wp-content/uploads/repo_items/legacy/partc/handout1.pdf

http://articles.extension.org/pages/28228/tips-for-child-care-providers-to-communicate-with-parents-their-concerns-about-a-childs-development

On the WWW

The Genetic and Rare Diseases Information Center (GARD) provides a wealth of useful resources including links to help connect individuals and families with rare medical conditions. These connections can be powerful and insightful. By connecting with others experiencing similar conditions they can understand how the condition might affect their lives now and in the future and experience support from others familiar with their unique situation. There are condition-specific groups as well as umbrella or alliance groups. Learn more about these supports and more at the GARD website. https://rarediseases.info.nih.gov/about-gard

Or link directly to support for patients and families at: https://rarediseases.info.nih.gov/guides/pages/120/support-for-patients-and-families

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.