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Newborn Screening Systems—The Complete Perspective: Interview with Bradley McPherson, PhD

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Douglas L. Beck, AuD, speaks with Dr. McPherson about his new book—*Newborn Screening Systems: The Complete Perspective*—as well as medical and genetic screenings, and more.

Academy: Good Morning, Brad. Thanks for taking the time to speak with me.

McPherson: Hi, Doug. My pleasure.

Academy: Brad, you have a rather unique professional appointment. If you don't mind, let's start with a brief discussion of your position?

McPherson: Sure thing. I was a senior lecturer in the department of speech pathology and audiology at the University of Queensland in Australia, but I am now an associate professor and the head of the division of Speech and Hearing Sciences and Director of the Center for Communication Disorders at the University of Hong Kong. Originally, I was just going to sign on and live here for a year or two, but I've been here since 1996. It is an intriguing place.

Academy: Hong Kong is quite a bit out of mainstream audiology! How many audiology programs are there in China?

McPherson: Not many, about three or four.

Academy: And do you speak any of the Chinese languages?

McPherson: Not really. I know a few words here and there, but all the courses are taught in English, and that's pretty common across university programs here in Hong Kong.

Academy: Wow. That's fascinating...and I'd love to hear more about that. Nonetheless—we really ought to discuss your new book *Newborn Screening Systems—The Complete Perspective* by Carlie J. Driscoll (University of Queensland, Australia) and you. The first thing that struck me when going through the book was that it was about newborn screenings in general—not just hearing screening issues and protocols.

McPherson: Right. And that does make it unique. I believe there are no other books out there that address all manner of newborn screenings, from screening principles, economics, ethics, newborn diseases and disorders, developmental disorders, genetic syndromes, and congenital malformations, as well as pediatric exams, the newborn dried bloodspot screening, genetics, communicating with parents and information management systems.

Academy: I have to admit, although I'm aware of APGAR scores, and of course I knew there were many other aspects of newborn screenings underway, I knew precious little about the other screenings.

McPherson: Right. We didn't know much about them either, and I think that's true for many professionals. That is, we get into a bit of a bubble focusing our energy and thoughts on our concerns and profession and what it is we do.

Academy: Yes, for me that's exactly right. When I hear the term "newborn screening" I think about ears, OAE, ABR and pass/fail criteria!

McPherson: Exactly—and even in a hospital setting, we rarely know what the other professionals are doing and what they're looking for, with respect to the same child we're concerned with—and so the book seemed like a good idea!

Academy: When one steps back and takes in the global picture, there are literally dozens of non-hearing related newborn screenings going on. Would you please define "newborn screening" for me, consistent with the more global view presented in the book?

McPherson: In chapter one, we talk about newborn/neonate screening as a systematic application of a test or inquiry within the first 28 days of life, to identify individuals at sufficient risk for a specific disorder, when those individuals would benefit from further investigation and/or preventative action for a given disorder.

Academy: And then if you would, can you please paraphrase the 10 principles of the more global newborn screenings?

McPherson: Yes, of course. And let me explain that these principles certainly do apply to hearing screenings, but they are also applicable to global screenings and are offered with consideration for more than the physical health of the child. That is, they consider political, economic, social, and health issues. So paraphrasing from the book, we have:

The condition we're screening for must be an important health problem, there should be an accepted treatment for the condition, there should be facilities (personnel, equipment, and environment) for diagnosis and treatment available, there should be a recognizable early and asymptomatic stage of the condition, suitable test(s) should be employed, the test should be relatively simple to perform, the natural history of the condition should be known, there should be an agreed policy with regard to who are

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the patients, the cost for the test and treatment should be appropriate and balanced, and the process should be ongoing, not a one-time event.

Academy: As you mentioned, these principles are written for the more global screening process, and in the book you offer examples of how each principle is demonstrated or realized in the real world, and of course, these same principles are applicable to newborn hearing screenings. The book addresses reliability and validity, sensitivity, and specificity, pass/fail criteria, false negatives and false positives and all the statistical probabilities that impact these screenings and their protocols.

McPherson: Yes, and it's important to understand that screening doesn't really, and shouldn't, exist in isolation. That is, newborn screening is related to all the steps in the program from identification and referral to confirmation and treatment.

Academy: And, of course, there are deeper concerns, too, such as ethical considerations.

McPherson: Undoubtedly. Rod Beattie contributed a very thoughtful and thought-provoking chapter on the internal and external realities and ethics of newborn hearing screening (NHS). Among many other issues and topics, he asks, "Does NHS respect the decision making autonomy of the parents, is the health concern more about beneficence (i.e., doing good) or paternalism, can components of NHS cause harm, and what about benefits and burdens to individuals and the larger society secondary to NHS?" For instance, when we screen and identify a child as hearing impaired, it may change and/or alter the parent's perception of their child—before they even get to know the child. And maybe that offers a benefit, but maybe that presents harm, too. Then, of course, one must consider what happens to the family that experiences a false positive or a false negative screening result.

Academy: Which gets us to the issue of "pass-versus-refer" rather than "pass-versus-fail."

McPherson: Right, and again, those words have internal meanings to the moms and dads and we cannot know what they're thinking when we use any of these terms. Therefore, we have an ethical obligation to communicate in such a way as to not cause harm to the individual or their families, while accomplishing our professional goals, too.

Academy: I know we cannot possibly address each newborn disease and disorder screening that occurs, but can you break them down into general categories for me?

McPherson: Sure. In general, newborn screenings include endocrine disorders, amino acidemias, organic acidemias, galactosemias, fatty acid oxidation disorders, hemoglobin disorders, and genetic and developmental disorders. And, of course, there are medical history and family histories, as well as the physical exam and more, and as you can imagine the newborn screenings vary state by state and country by country and they change, evolve, and improve over time. It's interesting to note that although some disorders are relatively rare, in general, congenital anomalies are present in some 9 percent of all infants, although some may not be apparent during the screening itself. So, the take-home lesson is that the screening is specific to that moment in time.

Academy: Yes, and, that, too, has direct application to hearing. In other words, a child can pass a newborn screening and then develop significant hearing loss in their first few months or years of life, such as might occur with cytomegalovirus.

McPherson: Exactly. And perhaps that gets us to the 2007 consensus statement from the JCIH on Universal Newborn Hearing Screening, which addresses the principles for an effective Early Hearing Detection and Intervention (EHDI) Program.

Summarizing, they said that

- All infants should be screened before one month of age;
- All infants who do not pass the initial or follow-up screening should receive audiologic and medical evaluations prior to three months of age;
- All infants with confirmed permanent hearing loss should receive intervention before six months of age;
- The EHDI program should be family-centered;
- Children should have immediate access to high-quality technology such as hearing aids, cochlear implants and assistive listening devices;
- Ongoing monitoring should occur in the "medical home";
- Appropriate interdisciplinary professional intervention should occur; and
- Systems should be designed to interface with electronic health records.

Academy: Thanks, Brad. Newborn screenings is clearly an epic topic and the book is quite fascinating and engaging. Although we could spend hours and hours on this, I know we have to end this discussion soon. Further, as many of us are familiar with audiologic screenings and we have access to those protocols, let's address one more non-audiology issue, such as the amount of information derived from the heel prick, also referred to as the "Newborn Dried Bloodspot Screening." What can you tell me about that?

McPherson: Well, simply, the dried bloodspot screening is indeed just a heel-prick and then a drop or two of blood is evaluated for biochemical markers, rather than disease itself. The test has been around for more than 50 years and the whole process is based on mass spectrometry, which continues to improve and expand. At this time, bloodspot screening is evaluating more than 50 congenital conditions, which can be detected—so it's a very important and pivotal analysis.

Academy: Thanks, Brad. I really learned quite a bit from your book, and it's been a pleasure speaking with you.

McPherson: Thanks, Doug. I enjoyed it, too.

Bradley McPherson, PhD, is an audiologist and co-author of the book Newborn Screening Systems – The Complete Perspective, published by Plural Publishing.

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