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INSIGHTS

Family reflections: kernicterus—diagnosis makes a world of difference

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Shortly after her birth in 2002, my youngest daughter, Lexi, developed newborn jaundice. Her doctor brushed off my concerns of Lexi's deepening yellow color and extreme lethargy. Her bilirubin was never measured. After 3 weeks, when we discovered her thyroid wasn't functioning and she began taking Synthroid, the jaundice finally cleared up. We had no idea jaundice could cause any long-term problems and frankly, as time passed, we didn't even connect the prolonged jaundice with the motor milestone delays that plagued her. In her first year, Lexi failed to meet a single motor milestone, but despite her motor delay it was becoming clear that she was incredibly bright. If I held her in my lap and supported her arm, she could drag magnetic letters around an old cookie sheet and spell. By her first birthday, she couldn't sit up, roll over, or babble, but she could read, spell, and properly use literally hundreds of words, including the homophones *their*, *there*, and *they're*. She was brilliant, but still undiagnosed. At 18 months old, she had a magnetic resonance imaging, which showed the defining injury that is kernicterus—a bilateral T2 hyperintensity of the globus pallidus—but even that didn't lead to a diagnosis until we found our way to Dr Steven Shapiro, MD, who at the time was in the Department of Neurology at the Medical College of Virginia in Richmond, VA.

Dr Shapiro teased apart her medical records. He showed us how she had the unambiguous injury to her globus pallidus (with devastating functional consequences). He showed us how her floppy, toneless neck, her twisting dystonic limbs and trunk, and her frail stained baby teeth all pointed to a diagnosis of kernicterus. As a medical writer, I had suspected that she had the motor predominant version of kernicterus, and I knew it wasn't the news any parent wanted to hear, but oddly, there was a sense of peace. It was a terrible, grim diagnosis, but it was a path forward. It meant a lifetime of disability and new questions on the daily, but it was also an answer.

Since then, as President of Parents of Infants and Children with Kernicterus, the only nonprofit organization dedicated to families affected by kernicterus, I have watched hundreds of families walk the path we walked. Most know early on what happened and that their baby has kernicterus, but many others like Lexi, aren't diagnosed right out of the gate. These families take their baby, toddler, grade-schooler, and sometimes even teenager from hospital to hospital searching for a diagnosis that doctors are either unable or unwilling to give. "It doesn't matter," or "What difference does it make if it's kernicterus?" the specialists tell families over and over. "It's not like we can give them a pill that will fix everything."

But what we as parents want doctors to understand is that a diagnosis does matter. In fact, it makes a world of difference.

A diagnosis matters because it gives families and these young patients an understanding of themselves. A diagnosis tells patients, their families, and the therapists and doctors who treat

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them that those muscle and hearing abnormalities, along with a host of other related issues, are *real* and explainable within the context of their condition. A diagnosis matters because it gives these children and parents a community—a loving, inclusive group of other families and children who have walked this same path and understand not only the heartache of a preventable condition but also the celebration for even the smallest physical achievement. A diagnosis matters and is imperative for these children's education. Kernicterus is not associated with cognitive impairment, and many of these children, like Lexi, are incredibly bright. They deserve an education that expects them to excel intellectually and provides the support for their bright minds to shine past the physical challenges.

Finally, and arguably most importantly, a diagnosis matters because there is strength in numbers. Our community is constantly told, "Kernicterus doesn't happen anymore." Doctors sometimes look at our children sitting right in front of them in their wheelchairs, with their cochlear implants, and tell them that kernicterus doesn't happen anymore. Diagnosis matters because it means our kids get to be counted. To be acknowledged. A diagnosis says *yes*, this preventable condition *is* still happening. It means that researchers will continue looking into prevention and treatment and that funding opportunities will be available for their valuable research. It means attention can be put towards prevention, which hopefully means fewer families will need to walk the path of Lexi and so many more before her.

For all these reasons, I, mom of Lexi, along with Parents of Infants and Children with Kernicterus and our kernicterus community, am ecstatic about the publication of "Kernicterus Spectrum Disorders Diagnostic Toolkit: validation using retrospective chart review." This important paper finally puts into physicians' hands the tool they need to make a retrospective diagnosis of Kernicterus Spectrum Disorders, potentially leading families to answers for what feels like an unsolvable enigma. It means children can receive proper treatment, and education and families can find support and community. It means our kernicterus community can finally be counted.

Every child with a disabling condition deserves a diagnosis. Many will never get one. I, for one, will never take for granted the peace diagnosis brings and am forever grateful for the physicians and researchers who have made this possible.

—Susan Haas is the President of Parents of Infants and Children with Kernicterus and co-author of *The Year of the Buttered Cat: a mostly true story* (Penelope Editions Press, 2021), which she co-wrote with her daughter Lexi detailing Lexi's experience with newborn jaundice.

COMPETING INTERESTS

The author declares no competing interests.

ADDITIONAL INFORMATION

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