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Genetics Detective Work Key to Difficult NICU Conversations

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I have always tried to teach medical students and residents this approach. When you are in the delivery room, newborn nursery, or neonatal intensive care unit (NICU), and you are evaluating a dysmorphic newborn, commit to doing your best to identify the syndrome – *before* the genetics team weighs in.

Joseph R. Hageman, MD, FAAP, NeoReviews Editorial Board

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"First, figure out what syndrome this baby has, and THEN call the geneticist!"

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When you see an anomaly in a newborn, be suspicious and look for other anomalies.

In the late 1970s and early 1980s, before there was fetal ultrasonography, fetal magnetic resonance imaging, chorionic villus sampling, or amniocentesis, I had the opportunity on multiple occasions to begin the evaluation of a dysmorphic newborn in the delivery room. As a pediatrician/neonatologist, I think you are also an amateur geneticist/dysmorphologist.

Honestly, when Drs. Kshama Shah and Alison DeRemigis expressed interest in writing a paper about X chromosomal disorders (3), I thought I could help as an amateur dysmorphologist!

I reviewed their early drafts and thought they did an excellent job of educating our clinical neonatologist audience. We then thought that Dr. Darrel Waggoner, our geneticist at University of Chicago, should review the paper to provide his expertise. He suggested that we should provide more specifics about the X chromosome – how it is inactivated and the effect of these changes on the phenotypes of the X chromosomal disorders.

And I have to admit, it has taken me rereading our paper with the help of Dr. Sudhir Sriram, who is a neonatologist who knows much more about genetics and dysmorphology, to refine this paper and make it more understandable and clinically relevant to our readers.

The result of this collaboration is "Unique Characteristics of the X Chromosome and Related Disorders."

As we have stated in the paper's introduction, the neonatologist often is the first clinician to explain the specifics of a syndrome to parents of a dysmorphic infant. Thus, it is important for neonatologists to be knowledgeable about genetic syndromes.

In situations where you are evaluating an undiagnosed dysmorphic newborn, I suggest referring to Smith's Recognizable Patterns of Human Malformation, which I have frequently used at the bedside during my evaluations of a dysmorphic newborn.

There are also clinically useful websites including http://www.omim.org/. Frequently, it takes me a number of times returning to the bedside with my resources to try to identify the infant's syndrome.

And please remember to review the prenatal course (Did the baby's mother have an amniocentesis? Did the quadruple screen reveal any abnormality? What about the fetal ultrasound? What is the ethnicity? What about the prenatal lab results?) to make certain that you have all of the pieces of the puzzle. It's important to perform a thorough physical examination, which may require repetition after you have reviewed your available resources.

After you have gathered all of your information and reviewed it with your colleagues, you can call your geneticist and proudly reveal your diagnosis.

Then, when the geneticist comes to examine your patient and says, "Joe, did you notice this clinodactyly, single-flexion crease, short stubby fingers, etc.," that you had not noticed, you can take a deep breath, learn how much more the specialist has observed … and remind yourself why you still need to call your genetics consultant in these clinical situations.

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