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Advances in the Interdisciplinary Care of Children with Klinefelter Syndrome

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ADVANCES IN PEDIATRICS

Advances in Pediatrics 63 (2016) 15-46

Advances in the Interdisciplinary Care of Children with Klinefelter Syndrome

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Keywords

Klinefelter syndrome • 47,XXY • Sex chromosome aneuploidy

Key points

- Klinefelter syndrome is a common but underdiagnosed genetic condition with significant phenotypic variability in childhood.
- The pediatrician needs to be aware of the increased risk for neurodevelopmental, psychological, and medical conditions that are associated with an additional X-chromosome.
- Over the next decade, we anticipate a sharp increase in diagnosis rates with advances in genetics, particularly prenatal and neonatal diagnoses.

K linefelter syndrome (KS) is a common genetic disorder characterized by an additional X-chromosome in male individuals leading to a karyotype of 47,XXY. The clinical syndrome was first described nearly 75 years ago in several male individuals with small testes, tall stature, gynecomastia, and azoospermia [1]. Our construct of what KS entails has greatly changed





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From the Department of Pediatrics, and Center for Bioethics and Humanities, State University of New York Upstate Medical University, Syracuse

The author has no conflicts of interest to disclose.

Address correspondence to Amy E. Caruso Brown, MD, MSc, MSCS, Center for Bioethics and Humanities, SUNY Upstate Medical University, 618 Irving Ave, Syracuse, NY 13210 (e-mail: brownamy@upstate.edu).

ACADEMIC PEDIATRICS 2017;17:343-344

MY NEWEST PATIENT was Michael (not his real name), a 16-year-old boy with a self-assured politeness that had charmed the nurses and impressed the chaplain. When I entered the room, after a brief knock and even briefer pause—more ritual than functional—he was sitting on the edge of his bed with his back to the door. His parents and sister formed a tight circle around him, their heads bowed in prayer. Only his father looked up and I gestured to him to continue. When they were finished, I pulled up a chair to talk about sperm banking.

A day earlier, Michael had presented to the emergency department complaining of difficulty breathing. A chest x-ray followed quickly by a computed tomography scan identified a large mediastinal mass, and he was admitted to our pediatric intensive care unit. His history of several months of fatigue, weight loss, and night sweats suggested Hodgkin disease. Impressive cervical lymphadenopathy had made an urgent biopsy under local anesthesia feasible. and treatment, but also how they can help families the most. The idea of asking families to put aside hope and fear and be eminently practical—to plan for a future that has abruptly become tenuous—can appear bizarre and irrational, or even insensitive.

As a pediatric oncologist, I sometimes think my most important role is to help families understand that their child's diagnosis is not the end of their lives—that, in most cases, there truly is a day after tomorrow. Nothing does that quite as singularly as the conversation regarding fertility preservation. I needed to encourage my patient to be able to imagine not just one, but many, possible futures, to help him understand what it means to keep a door open, while at the same time, validating his current feelings and respecting his choices and nascent autonomy.

In a perfect world, this would be a conversation held only after painstakingly building a rapport between physician, patient, and family, including a thorough under-



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