This case begins with a prenatal consultation. During the second trimester of the pregnancy of a 27-year-old G1P1 woman, a supra-renal mass was discovered by ultrasoundography. The subsequent pregnancy, labor, and delivery were normal. The family history is unremarkable.

Robert Listernick, MD, moderator: How routine is ultrasonography during pregnancy in the United States?

Susan Gerber, MD, MPH, maternal-fetal medicine specialist: The overwhelming majority of women in urban areas who have prenatal care receive an ultrasound at approximately 20 weeks of gestation for an anatomic survey.

Dr. Listernick: To what end?

Dr. Gerber: The value is twofold. First, some patients would choose to terminate a pregnancy if they found out that the fetus had a life-threatening condition or one that was going to significantly impair quality of life. Second, there are good data that there is a better outcome for fetuses that have a prenatally diagnosed anomaly if they are delivered at a tertiary care referral center rather than at a community hospital.

Dr. Listernick: Are there data on the sensitivity and specificity of ultrasound screening for finding significant structural abnormalities?

Dr. Gerber: In the best hands, although this still varies among institutions, the sensitivity is approximately 80% for the detection of significant cardiac, central nervous system, or gastrointestinal anomalies. We’re more likely to miss minor anomalies such as small atrial septal defects.

Dr. Listernick: Are there high-risk populations of women who should definitely undergo ultrasound screening?

Dr. Gerber: Women who are obese, diabetic, have advanced maternal age, or who take a variety of potential teratogens such as antiepileptic medications all should undergo screening since they are at greater risk of having babies with structural abnormalities.

Dr. Listernick: Can the ultrasound screening be “overly sensitive”?

Dr. Gerber: Women who are obese, diabetic, have advanced maternal age, or who take a variety of potential teratogens such as antiepileptic medications all should undergo screening since they are at greater risk of having babies with structural abnormalities.

Dr. Listernick: Assumptions with parents who wish 100% certainty, which we can’t provide.

Dr. Listernick: Let’s look at the post-natal ultrasound.

James Donaldson, MD, pediatric radiologist: This study was performed on the day of birth. There’s a solid mass sitting above the left kidney with a significant amount of vascular flow within it, as seen by color Doppler imaging. Although I can’t be sure, this mass looks discreet from a normal-appearing adrenal gland. A follow-up study performed 2 months later looks virtually the same; the mass hasn’t grown.

Dr. Listernick: Differential diagnosis?

Dr. Donaldson: That depends upon how the study is interpreted. If you feel that it might be arising from the adrenal gland, then neuroblastoma rises high on the list. If you believe that it’s separate from the adrenal gland, by far the most likely diagnosis is an extrapulmonary sequestration. I can’t be sure from where its blood supply is coming. A sequestration may have its blood supply from the abdominal aorta, the celiac artery, or the descending aorta.

Dr. Listernick: Assuming this mass represents a neuroblastoma for the moment, are they sometimes detected in utero?

David Walterhouse, MD, pediatric oncologist: Absolutely. However,
there has been a long-standing debate on how to approach them.

**Dr. Listernick:** There’s a lot of experience with mass newborn screening?

**Dr. Walterhouse:** Most neuroblastomas secrete catecholamines, vanillylmandelic acid (VMA) and homovanillic acid (HVA), which can be detected in the urine. Several countries, including Japan and Canada, have trialed newborn screening programs with the ultimate goal to prevent mortality from neuroblastoma in toddlers who present with high-risk metastatic neuroblastoma. Unfortunately, that wasn’t the end result. Instead, they identified higher rates of neuroblastoma than had been described previously. It turned out that they were identifying tumors whose ultimate fate was to regress that would never have been identified but for the newborn urine screening.

**Dr. Listernick:** How did they know prospectively that these tumors would regress?

**Dr. Walterhouse:** They didn’t. However, a recently published study looked prospectively at infants younger than 6 months of age who had an identified tumor discovered that was smaller than 3 cm in its greatest dimension. These tumors were observed without any intervention, including biopsy. The patients were offered immediate surgery or observation. At the beginning of the study, it was agreed that in those who opted for observation, the tumor would be removed if it increased in volume by 50%. There was extremely close follow-up with ultrasonography and urine VMA and HVA. Of the 88 patients, only four decided upon surgery. As they were followed, most of the tumors became smaller and disappeared. Some that were removed were not neuroblastoma. Most importantly, there were no deaths in the study patients.

**Dr. Listernick:** So is any country performing neonatal neuroblastoma screening?

**Dr. Walterhouse:** I believe they have all stopped because a great deal of anxiety was produced without any survival benefit.

**Dr. Listernick:** If you had been involved with this patient, would you have performed urine screening for neuroblastoma?

**Dr. Walterhouse:** If there was question as to whether this mass was of adrenal origin, then that would have been reasonable. However, in the study I cited, only approximately 20% of the patients with neuroblastoma had elevated urine levels of HVA or VMA.

**Dr. Listernick:** When the ultrasound was reviewed here, it was clear that the mass was discrete from the adrenal gland, which changes the differential diagnosis. But let’s assume for the sake of discussion that neuroblastoma was a possibility and watchful waiting was a reasonable approach. Essentially, you’re telling a family, “your child may have cancer but that we want to keep a close eye on it. Alternatively, we can remove it.” What do you say to the parents when they ask, “What would you do if this were your child, doc?” I’ll start by saying that I never answer that question. I’d tell them it’s a very personal decision that involves both the medical facts as well as their personal beliefs and that I can’t make that decision for them.

**Dr. Donaldson:** I don’t necessarily agree. I think it’s a fair question. The family is asking me based on my education, background, and understanding of the disease process to help them make a decision.

**Joel Frader, MD, pediatric ethicist:** There are really two questions here. First, whether we should make recommendations. I think most of us believe that we should make recommendations based on our knowledge and our clinical experience. However,
the question, “What would we do if this were our child?” is somewhat different and much more complicated. When I am asked that question, I first try to get a better understanding from the family about what they’re really asking. On occasion, I find that what they are really concerned about are the risks and I try to get a sense of their “risk tolerance” in order to help. If they still pursue this line of questioning, my basic response to that is to say that 1) this is a very difficult question to answer, 2) I can’t put myself in your shoes without a better understanding of your beliefs and family support, but 3) given these caveats, here’s what I might do if I were in a similar situation. I don’t think that we should duck the question when families press us; I think we should try to answer it, but it’s a prolonged, difficult conversation.

Britt Allen, MD, pediatric chief resident: I agree with Joel, but I also think our response might differ depending upon the complexity of the medical question. For instance, if asked if I would give my own child any vaccination, I would certainly answer unequivocally “yes.” I would probably balk at answering the question in this particular situation.

Angira Patel, MD, pediatric cardiologist: This is a tension that we’ve been feeling in medicine over the past 20 years as we’ve transitioned from a somewhat paternalistic attitude to more patient autonomy. I feel that, at times, the pendulum has swung too far to patient autonomy, such that physicians will often say, “You can do this, you can do that, you can choose, etc.” I believe that there is a real danger of the patient or family feeling abandoned if this approach is applied too liberally. Sometimes, I think that it’s our responsibility to “step up to the plate” and say what we really think based on our experience.

Dr. Listernick: OK, we could continue this discussion for hours. Just for completeness, what would happen if this mass were removed and turned out to be neuroblastoma?

Dr. Walterhouse: We would classify the tumor as having favorable or unfavorable histology and determine prognostic factors based on its biology — degree of MYCN amplification, DNA ploidy of the tumor, and loss of heterozygosity at 1p and 11q. Most of the time in an infant, all of these factors are favorable and we give no further therapy.

Dr. Listernick: Great. However, once the radiology studies were examined closely here, it was felt that the mass was not arising from the adrenal gland. Thus, neuroblastoma was not considered likely and the preoperative diagnosis was extra-pulmonary sequestration. Why did it need to be removed?

Loren Berman, MD, pediatric surgeon: We were virtually 100% certain that the mass was an extrapulmonary sequestration. These masses have a definite risk of infection and there’s a debatable concern about future malignancy. It’s a lot easier to remove them before they have become infected. We performed a laparoscopic-assisted approach in which we first obtained a lot of exposure of the mass using the laparoscope and then made a small incision to remove the mass from the blood supply coming from the abdominal aorta. In this way, we were able to leave the child with a much smaller incision than would have been possible with a completely open procedure. It was indeed an extrapulmonary sequestration.

Dr. Listernick: Why do these form in the abdomen?

Dr. Berman: The theory is that pulmonary sequestrations form from accessory lung buds that develop from the ventral surface of the primitive foregut. Their blood supply is from the aorta. If the development goes awry, these “sequestered” areas may develop within the same pleura as the embryonic lung (intralobar sequestration) or outside of the pleural space. Rarely, they may communicate with the gastrointestinal tract and have blood supply from the abdominal aorta or celiac axis.

Dr. Listernick: Thank you, everyone.