

Rare Fetal Sacral Appendage Studied

BY DOUG BRUNK

FROM THE ANNUAL MEETING OF THE AMERICAN INSTITUTE OF ULTRASOUND IN MEDICINE

SAN DIEGO — A fetal sacral appendage carries a guarded prognosis, especially in the setting of associated anomalies, results from a single-center study demonstrated.

"Fetal sonography is still the primary mode for identifying the extension," Lori J. Dobson said during the meeting. "It is important to measure fetal growth and look at the umbilical cord Doppler [scans], as well as to survey for additional anomalies. We did show that it is important to do a fetal MRI as it can help identify additional anomalies and help determine the position of the spinal cord conus."

A fetal sacral appendage—also referred to as a fetal "tail"—is part of normal embryonic development, expected to resolve by the 8th week of gestation. While more than 30 published studies of prenatally diagnosed fetal sacral appendages have appeared in the medical literature, "the majority of cases are isolated, with no other structural anomalies reported," said Ms. Dobson, a certified genetic counselor in the advanced fetal care center at Children's Hospital Boston. "And in most cases the appendage had resolved by the second trimester."

She and her coinvestigators, some of whom performed fetal imaging, set out to describe the associated anomalies, etiology, and clinical outcome for a subset of the 3,641 patients evaluated at the hospital's advanced fetal care center over a 9-year period that ended in December 2009. Ms. Dobson reported that of the 3,641 patients, 7 fetuses (0.19%) had sacral appendages that ranged in size from 2.1 to 4.5 mm. The researchers assessed findings on fetal sonography and fetal MRI, results of

genetic testing, and clinical outcomes.

"We noted that in all seven cases there was an extension of the coccyx causing a protrusion or a tenting of the skin," she said. "Based on this, we propose the term sacrococcygeal extension to better describe the physical finding and its etiology. This is also a more patient-friendly term than fetal tail to use in counseling with parents."

The average gestational age at diagnosis was 19 weeks. "Because we are a tertiary care referral center, it is not uncommon for us to only see patients in the

VITALS

Major Finding: All seven fetuses with fetal sacral appendages had significant associated anomalies, and only one is surviving.

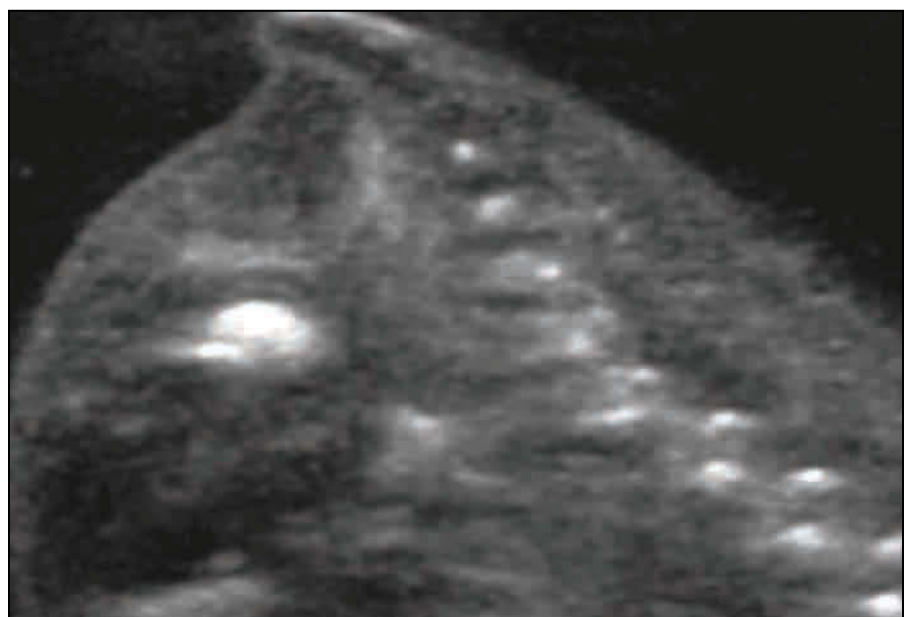
Data Source: A single-center study of seven fetuses with fetal sacral appendages evaluated over a 9-year period.

Disclosures: None was reported.

second trimester or after," she noted. "That may represent a bias in our sample. We did not have any cases that were diagnosed in the first or third trimester."

All seven of the fetuses with sacral appendages had significant associated anomalies including severe growth restriction and neurologic, spinal, craniofacial, cardiac, renal, and musculoskeletal abnormalities. There were two cases of trisomy 13 and one case of Pfeiffer syndrome (craniosynostosis). "Most of our cases had multiple organ systems involved," Ms. Dobson said.

She went on to report that three of the fetuses died in utero, two underwent elective termination, and one fetus with trisomy 13 was delivered at 36 weeks' gestation and died on the first day of life. The remaining case carried to term, and the infant is doing well at 5 months old, but does have a sacral appendage with a tethered spinal cord and an abnormal distal spine. ■



This sagittal view shows a fetal sacrococcygeal extension on ultrasound at 19 weeks', 4 days' gestation.

DRUGS, PREGNANCY, AND LACTATION

Seafood Safety

Although fish are a healthy source of protein that is low in saturated fat and contains omega-3 fatty acids, which may be important for fetal brain development, fish contain methylmercury, which is known to exert adverse effects on fetal brain development. The amount of methylmercury in fish varies widely depending on the type of fish. The Food and Drug Administration and Environmental Protection Agency recommend that pregnant women not eat swordfish, shark, king mackerel, or tilefish, and that they limit their fish and shellfish intake to no more than 12 ounces a week (two average meals) of a variety of fish and shellfish that are lower in mercury.

What has not been clearly defined is the lowest concentration of maternal mercury that is associated with observable neurodevelopmental effects in the unborn child—a question that has been addressed by many scientists in many countries. In two recent studies, we provided new evidence that support testing maternal hair for mercury levels in some groups of pregnant women and suggests that analyzing a woman's hair mercury content before she becomes pregnant might be useful.

In an attempt to define the lowest observable adverse effect level (LOAEL) of mercury, our group conducted a systematic literature review of 48 studies on the effects of prenatal exposure to mercury on the fetus, which used mercury levels in maternal hair samples to estimate prenatal methylmercury exposure. In the studies, adverse neurodevelopmental abnormalities were found at a range of maternal mercury levels based on results of hair sample analyses. We decided that the precautionary principle should prevail: There were variabilities among the studies, but we concluded that the lowest level—0.3 mcg/g—at which adverse events were documented in at least one or two studies should be the level that should not be exceeded (*Ther. Drug Monit.* 2009;31:670-82).

We applied these consensus results to the clinical setting, which involved analyzing hair mercury levels and associating those levels with the amount of fish in the diet in 23 Japanese women living in Toronto who ate fish every day, 22 pregnant women who ate fish regularly and called Motherisk with concerns, and 20 Canadian women who had no concerns about excessive consumption of fish. All the Japanese women, two-thirds of the women who called Motherisk, and 15% of the Canadian women who were not pregnant had levels that exceeded 0.3 mcg/g.

The median level of mercury in the hair samples of the Japanese women was 1.7 mcg/g, significantly higher than the median level among those women who called Motherisk (0.4 mcg/g) and those in the general population (0.2 mcg/g). The study was published online in the *Journal of Pediatrics* (2010 March 24 [doi:10.1016/j.jpeds.2010.01.020]).

The median number of servings of fish per month was also significantly different among the three groups, with

10 servings a month among the Japanese women, 4 servings a month among the women who called Motherisk, and 3 servings a month among the Canadian women. As in other studies, the hair mercury level correlated with the amount of fish consumed. But the data suggested that in only half of the cases could the variability in hair mercury be

accounted for by the amount of mercury estimated in the fish the women consumed: Even among some of the women whose fish intake fell within the FDA/EPA recommendations for pregnant women, levels exceeded 0.3 mcg/g, which in the previous study, was assigned as the LOAEL.

Therefore, although women who follow the FDA/EPA recommendations are probably under the limit, our data show this is not universally true. The study had some limitations, but the results still indicate that following these guidelines may not be adequate to prevent the risk of exposure to levels exceeding the minimum associated with neurodevelopmental effects.

These findings are the basis of a new initiative at Motherisk, where women who call with concerns about fish intake are counseled about the recommended amount during pregnancy but are also offered the hair test. The test can be used to reassure these women that their levels are below 0.3 mcg/g.

The FDA/EPA recommendations are based on the type of fish and amount eaten per week. The amount of mercury in fish is widely variable, however, so we recommend a hair mercury analysis for those women who eat more than the recommended amount of fish. Mercury hair testing is not widely available yet, but it is performed at most university medical centers.

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