Fetal spinal dysraphism: The conus medullaris and the spectrum of neural tube defects

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Objective
To assess conus medullaris involvement and appearances in both OSD and CSD. To observe prenatal US, MRI, and clinical features seen in fetuses with closed spinal dysraphism (CSD).

Methods
Prenatal imaging and postnatal clinical records were reviewed to identify fetuses and children with clinical, radiological and/or surgical evidence of OSD and CSD between 1997 and 2012.

Results
328 cases of OSD (excluding anencephaly) were identified correctly with prenatal imaging. In all OSB cases, the conus was abnormal. There were 35 cases of CSD identified. The conus was abnormal in all detected lumbosacral lesions. In CSD, MSAFP was negative in 10 cases and elevated in 2. AFAFP and ACE were elevated in 1 case. US did not identify 11 CSD cases before 24 weeks: all were spinal lipomas. In one case of CSD there was evidence of mild Chiari II malformation and borderline ventriculomegaly on US (thoracic myelocystocele); no Chiari II malformation was identified in any other case and there was no ventriculomegaly. In 18/20 sonographically detected lumbosacral lesions, a low conus was seen (in 1 case, the conus was in normal position). Prenatal US findings among the 20 spinal lipomas included a cyst or mass (14 cases); vertebral dysraphic anomalies (14), fat within the spinal canal (11) and talipes (4). In 1 spinal lipoma, fat was not seen on fetal MRI (<24 weeks).

Conclusion
A low or non-visualized conus is a reliable finding in lumbosacral OSD and CSD. The prenatal diagnosis of CSD remains more challenging than OSD. MSAFP, AFAFP and ACE can be normal or elevated in CSD. Cysts, fatty masses, vertebral, and foot abnormalities are associated with both OSD and CSD. Spinal lipomas may not be appreciated on US and MRI, particularly before 24 weeks, due to a paucity of fetal body fat as well as lack of MR signal. Since inability to visualize the conus medullaris is a feature of both OSD and CSD, documentation of the conus on routine ultrasound and MRI screening views would enhance and confirm detection of both kinds of fetal lumbosacral NTD.