SCIENTIFIC ARTICLE

Prenatal Detection of Upper Limb Differences With Obstetric Ultrasound

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Purpose To determine the sensitivity, specificity, and predictive values of prenatal ultrasound detection of fetal upper extremity anomalies at a single tertiary care center in a large patient cohort. Our secondary purpose was to assess factors affecting prenatal detection including the presence of associated anomalies.

Methods We performed a retrospective review of prenatal ultrasound and postnatal clinical records from each pregnancy evaluated with a prenatal ultrasound at the Washington University Department of Obstetrics and Gynecology over a 20-year period. We searched for upper extremity anomaly diagnosis codes pre- and postnatally and correlated with clinical postnatal follow-up to determine prevalence, sensitivity, specificity, predictive values, and associated conditions.

Results A total of 100,856 pregnancies were evaluated by prenatal ultrasound, which included 843 fetuses diagnosed with a musculoskeletal anomaly (prevalence, 1 of 120) and 642 with an upper extremity anomaly (prevalence, 1 of 157). The postnatally confirmed sensitivity for prenatal ultrasound detection of an upper extremity anomaly was 42%. Sensitivity was lower in cases isolated to the upper extremity (25% vs 55%). Sensitivity was highest for conditions affecting the entire upper extremity (70%−100%) and lowest for those affecting the digits alone (4%−19%). Fetuses with limb reduction defects, radial longitudinal deficiency, phocomelia, arthrogryposis, abnormal hand positioning, and cleft hand had a higher likelihood of having an associated anomaly.

Conclusions At our tertiary referral center, there was a notable prevalence of upper extremity anomalies; however, the overall sensitivity for detecting them with prenatal ultrasound was low. This was disappointing given the value of prenatal identification of anomalies for parental counseling. Prenatal diagnosis of anomalies affecting the entire upper limb was more reliable than diagnosis of more distal anomalies. *(J Hand Surg Am. 2015;40(7):1310−1317.*

Type of study/level of evidence Diagnostic III.

Key words Congenital limb anomaly, prenatal detection, prenatal obstetric ultrasound, birth anomaly, gestation.

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Prenatal detection and diagnosis of upper limb anomalies provides an opportunity for parental counseling and may prompt earlier diagnosis and treatment of associated conditions. Ultrasound remains the primary modality for fetal evaluation,[1] and detection of fetal anomalies has improved substantially over the last several decades as a result of technological advances, improved resolution, standardization of prenatal ultrasound protocols, and training of diagnosticians.[2−7] Recent studies have shown the sensitivity

for detection of major fetal anomalies to be over 70%, although the detection of musculoskeletal anomalies remains lower, ranging from 18% to 40%.[6,8] Evaluation of the upper extremity, especially the small structures of the hand, can be difficult, with sensitivities between 20% and 30%.[2,7,9] Detection rates tend to be higher in tertiary care centers for high-risk patients and for fetuses with multiple anomalies.[8]

The aims of this study were to evaluate the performance of prenatal ultrasound detection of fetal upper limb defects at a single tertiary care center, to determine which upper limb defects were the most and least likely to be diagnosed prenatally, to understand the relative frequency of upper extremity anomalies, and to determine if upper extremity anomalies with concomitant associated conditions had higher detection rates.

MATERIALS AND METHODS

After approval by the human research protection office at Washington University School of Medicine, we performed a retrospective review of all pregnancies evaluated with a prenatal ultrasound at the Washington University Department of Obstetrics and Gynecology between January 1990 and January 2010. This is a tertiary referral center that provides care to both high- and low-risk pregnant women and performs both standard and specialized obstetric ultrasound examinations. Registered diagnostic medical sonographers with certification in obstetrics and gynecology performed the examinations, and maternal-fetal medicine specialists interpreted them. A dedicated nurse coordinator acquired postnatal clinical follow-up data on 94% of births, allowing for validation of prenatal ultrasound findings and identification of anomalies not diagnosed by ultrasound (false negatives).

The Department of Obstetrics and Gynecology database includes all pregnancies with prenatal or postnatal diagnoses of any anomaly. This database contains indications for the ultrasound, prenatal ultrasound fetal diagnostic codes and clinical notes from each examination, number of ultrasounds per pregnancy, gestational age at each ultrasound, outcome of each pregnancy, postnatal diagnostic codes and clinical notes, and all associated diagnoses for each fetus.

The database was queried for pre- and postnatal institutional fetal anomaly diagnostic codes for upper extremity conditions including major and minor skeletal anomalies, limb reduction defects, abnormal hand positioning, arthrogryposis, polydactyly, syndactyly, and amniotic bands. Arthrogryposis was defined as fetal joint flexion or extension contractures with rigidity involving more than one large joint (hip, knee, ankle, elbow, or wrist). Abnormal hand positioning was defined as clenched hands or overlapping digits. Obstetricians use the diagnosis of abnormal hand positioning as a potential marker for the presence of nonorthopedic malformations rather than as an independent diagnosis potentially requiring treatment. Limb reduction defect was defined by shortening and/or absence of the bone(s) of one or more extremities. When possible, we supplemented the database with a review of the medical records at our institution to further refine and subcategorize diagnoses such as ulnar and radial longitudinal deficiency, cleft hand, symbrachydactyly, and phocomelia.

We identified each prenatal diagnosis as either a true positive or a false-negative after reviewing postnatal clinical findings. The true-negatives group included the 100,013 live births (LB) during the study period that had a negative ultrasound for upper extremity anomalies and a postnatal examination with no coded diagnosis of an upper extremity anomaly. We then calculated prevalence, sensitivity (with confidence intervals [CI]), specificity, and predictive values for each diagnosis.

We categorized the indications for prenatal ultrasound into 2 groups. When patients were referred for prenatal ultrasound evaluation of a known or suspected fetal musculoskeletal malformation, they were labeled high risk. Examinations performed for other indications were classified as other. Age at diagnosis was identified and categorized into either before or after 22 weeks of gestation, because beyond this age, termination of pregnancy (TOP) options are limited. We divided pregnancy outcomes into LB, stillbirth, TOP, and neonatal death. Findings were classified as isolated if no other anomaly was present on postnatal clinical follow-up or as associated with other systemic anomalies on postnatal clinical examination. These other systemic anomalies were categorized as central nervous system, facial, nuchal, cardiothoracic, abdominal, renal, aneuploidy, or specific syndrome diagnoses.

RESULTS

There were 100,856 pregnancies evaluated with an obstetric ultrasound at a gestational age greater than or equal to 13 weeks during the study period. Of these, 350 (0.3%) of the pregnancies were considered high risk, the situation in which a musculoskeletal anomaly was suspected upon referral to our center, and 100,506 (99.7%) were referred to our institution for an indication for assessment other than a suspected musculoskeletal