

## ORIGINAL ARTICLE

# The utility of ultrasound for the detection of fetal limb abnormalities – a 20-year single-center experience

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## ABSTRACT

**Objective** The standard obstetric ultrasound examination includes documentation of arms and legs. The purpose of this study is to review the efficacy of ultrasound for the detection of limb anomalies, the type and incidence of associated malformations and pregnancy outcomes.

**Method** All cases of polydactyly (POD), abnormal hand position (AHP), limb reduction defects (LRD) and arthrogryposis (ART) scanned in our Unit between 1990 and 2010 were identified. Cases were categorized as isolated (ISO) or non-isolated (NISO). The accuracy of prenatal diagnosis, type and incidence of associated malformations and aneuploidy and pregnancy outcomes were determined.

**Results** Most cases were NISO. The sensitivity of ultrasound was 19.1% for POD, 76.0% for AHP, 76.0% for LRD involving the long bones and 81.3% for ART. Cardiothoracic and facial defects were the most common accompanying malformations. The risk for aneuploidy ranged from 3.6% for POD to 47.2% for AHP. The live birth rate ranged from 85.5% for POD to 24.5% for AHP.

**Conclusion** While imaging of the arms and legs allows detection of most cases of AHP, LRD involving the limbs and ART, a probable minimum of 20% to 25% of cases will escape prenatal diagnosis. Identification of these defects should prompt an extended anatomic survey and consideration of invasive testing for aneuploidy. © 2014 The Authors. *Prenatal Diagnosis* published by John Wiley & Sons Ltd.

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## INTRODUCTION

Congenital limb abnormalities have a live birth (LB) prevalence of approximately 1 in 1600.<sup>1</sup> The prenatal detection of such malformations is important for parental education, counseling, initiating appropriate planning for postnatal evaluation and treatment and ruling out associated malformations, which may alter the management of the pregnancy. The recent Executive Summary of a Joint Eunice Kennedy Shriver National Institute of Child Health and Human Development, Society for Maternal-Fetal Medicine, American Institute of Ultrasound in Medicine, American College of Obstetricians and Gynecologists, American College of Radiology, Society for Pediatric Radiology, and Society of Radiologists in Ultrasound Fetal Imaging Workshop recommends documentation of the arms and legs as a component of the standard fetal examination at 18 to 20 weeks of gestation.<sup>2</sup> However, the utility of visualizing these structures in excluding anomalies of the extremities is variable.<sup>3</sup> The purpose of this study was to review the diagnostic accuracy of obstetric ultrasound at a tertiary referral center in the United

States for the detection of fetal limb abnormalities, associated malformations and outcomes.

## METHODS

This is a retrospective review of all patients who underwent obstetric ultrasound examination in the Department of Obstetrics & Gynecology at Washington University School of Medicine between January 1990 and January 2010. Our Unit is a tertiary referral center for standard as well as specialized ultrasound examinations. All prenatal and postnatal ultrasound and pregnancy outcome data are stored in a computerized database, and a dedicated nurse coordinator attempts to obtain neonatal follow-up on all patients having prenatal ultrasound. This allows confirmation of prenatal ultrasound findings as well as postnatal determination of conditions not detected prenatally and pregnancy outcomes. All examinations were performed by registered diagnostic medical sonographers with certification in obstetrics and gynecology. All examinations were interpreted by maternal-

fetal medicine sub-specialists or board certified obstetrician-gynecologists with extensive experience in fetal imaging.

In this investigation, women whose fetuses were identified as having polydactyly (POD), arthrogryposis (ART), limb reduction defects (LRD) or abnormal hand positioning (AHP) prenatally and/or postnatally were identified in the computerized data base. ART was defined as fetal joint contractures and rigidity. AHP was defined as clenched hands or overlapping digits. For each condition, the indication for the initial ultrasound, the accuracy of prenatal diagnosis, the pregnancy outcome and associated malformations were reviewed. The indication for examination was classified as either evaluation of a known or suspected malformation at the time of diagnosis or 'Other'. Cases with a known or suspected malformation were often referred for suspicion of a major malformation, which was non-skeletal. However, the fact that these fetuses were scanned in follow-up to an abnormal screening examination may have contributed to the diagnosis of our conditions as ancillary findings. Cases in the 'Other' category were typically referred for gestational age assignment, standard anatomic survey, fetal growth or maternal medical or obstetric conditions.

Prenatal diagnosis was classified as (1) true positive (TP) if the prenatal findings were confirmed on examination of the infant, (2) unconfirmed positive (UP) if the prenatal findings were unable to be confirmed, usually because of pregnancy termination with no autopsy, (3) false positive (FP) if the prenatal diagnosis was not confirmed postnatally and (4) false negative (FN) if the limb abnormalities were detected postnatally, but not prenatally. Outcomes were classified as live-birth (LB), termination of pregnancy (TOP), stillbirth (STB) and neonatal death (NND). Associated malformations were categorized as follows: central nervous system, facial, nuchal, cardiothoracic, abdominal and renal. Exclusion criteria included lack of known pregnancy outcome. This study was approved by the Human Research Protection Office at Washington University School of Medicine.

RESULTS

During the study period, there were 100 013 LBs that were scanned at greater than 13 weeks with known neonatal outcomes and no identification of a skeletal anomaly. The findings are summarized in Tables 1–7. The number of cases

Table 1 The types and numbers of fetal limb anomalies

Limb anomalies: isolated (ISO) versus non-isolated (NISO)			
	N	ISO	NISO
POD	330	250(75.8%)	80(24.2%)
AHP	110	12(10.9%)	98(89.1%)
LRD	120	50(41.7%)	70(58.3%)
Limb	75	29(38.7%)	46(61.3%)
Digit	38	21(55.3%)	17(44.7%)
Unsp	7	0	7(100%)
ART	36	12(33.3%)	24(66.6%)

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; Unsp, unspecified; ART, arthrogryposis.

Table 2 The gestational age at diagnosis of fetal limb anomalies

Gestational age at diagnosis (weeks)			
	N	≤22	>22
POD	330	47(14.2%)	283(85.8%)
AHP	110	63(57.3%)	47(42.7%)
LRD	120	78(65.0%)	42(35.0%)
Limb	75	46(61.3%)	29(38.7%)
Digit	38	27(71.1%)	11(28.9%)
Unsp	7	5(71.4%)	2(28.6%)
ART	36	21(58.3%)	15(41.7%)

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis; Unsp, unspecified.

of POD, AHP, LRD and ART is shown in Table 1. The prevalence of these conditions was as follows: POD, 1/300; AHP, 1/910; LRD, 1/835; and ART, 1/2780. Of all LRD, 62% involved one or more limbs and 32% involved the digits. Six percent were not specified. Each condition was classified as isolated (ISO) or non-isolated (NISO) depending on the presence or absence of associated malformations on postnatal examination. POD and LRD of the digits were the only conditions more likely to be ISO; the NISO rate for the other conditions ranged from 58% for LRD to 89% for AHP (Table 1).

The gestational age at diagnosis in our Unit was stratified to 22 weeks or less and greater than 22 weeks. The percentage of cases detected at a gestational age when all reproductive options would be available at our center was similar for AHP, LRD and ART and ranged from 57% to 65% (Table 2). Diagnosis of POD was limited to 15%.

As illustrated in Table 3, other than POD, which was usually ISO and not suspected prior to diagnosis in our Unit, 44% to 53% of the total number of cases of LRD, AHP and ART were detected or confirmed following referral for a suspected malformation. However, when only ISO cases are considered, 62% to 75% were unsuspected prior to our examination.

The accuracy and screening efficiency of ultrasound is depicted in Table 4. POD, even when NISO, was least likely to be detected and had a much higher FN rate. All conditions, except ART, were less likely to be diagnosed when ISO. Combined TP and UP rates for ISO POD, AHP and LRD were 17.6%, 50.0% and 52.0%, respectively. Corresponding TP and UP rates for NISO POD, AHP and LRD were 22.5%, 68.4% and 67.1%. Overall, combined TP and UP rates were 18.8% for POD, 66.4% for AHP, 60.8% for LRD and 72.2% for ART. ART was reliably diagnosed even when ISO with a TP and UP rate of 91.2%. FP rates were low in all groups but more likely in NISO cases, which may reflect a tendency to over-diagnose these conditions in the setting of other malformations. The overall sensitivity and specificity of ultrasound for the detection of these disorders is seen in Table 5. The sensitivity was low for POD and LRD involving only the digits (19.1% and 28.9%, respectively) but in a clinically useful range for ART (81.3%), AHP (76.0%) and LRD involving the limbs (76.0%). Corresponding positive predictive values (PPVs) ranged from 84% for AHP to 97% for LRD. The specificity and negative PV (NPV) exceeded 99% for all categories. The effect of time during

Table 3 The referral indication for ultrasound examination

Indication for examination			
	N	Known or suspected malformation	Other
POD	330	71(21.5%)	259(78.5%)
ISO	250	21(8.4%)	229(91.6%)
NISO	80	50(62.5%)	30(37.5%)
AHP	110	58(52.7%)	52(47.3%)
ISO	12	3(25.0%)	9(75.0%)
NISO	98	55(56.1%)	43(43.9%)
LRD	120	53(44.2%)	67(55.8%)
ISO	50	19(38.0%)	31(62.0%)
NISO	70	34(48.6%)	36(51.4%)
ART	36	17(47.2%)	19(52.8%)
ISO	12	4(25.0%)	8(75.0%)
NISO	24	13(54.2%)	11(45.8%)

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis; ISO, isolated; NISO, non-isolated.

the study period, gestational age and indication for the examination on the sensitivity and PPV of ultrasound for the detection of fetal limb abnormalities are depicted in Tables 6 and 7. AHP and ART were more likely to be detected in the second half of the study than in the first, although there was no difference for LRD, and POD was less likely to be diagnosed. Detection of AHP, LRD and ART was similar whether the examination was performed before or after 22 weeks of gestation. POD was much less likely to be identified after 22 weeks. The likelihood of correctly identifying a limb abnormality was enhanced when a malformation of any type was suspected at the time of referral to our center.

Corresponding PPVs are depicted in Table 7 and range from 80% for ART to greater than 97% for LRD, regardless of time

Table 4 The accuracy of ultrasound prenatal diagnosis of limb anomalies

Prenatal diagnosis					
	N	TP	UP	FP	FN
POD	330	51(15.5%)	11(3.3%)	5(1.5%)	263(79.7%)
ISO	250	34(13.6%)	10(4.0%)	1(0.4%)	205(82.0%)
NISO	80	17(21.3%)	1(1.3%)	4(5.0%)	58(72.5%)
AHP	110	32(29.1%)	41(37.3%)	14(12.7%)	23(20.9%)
ISO	12	2(16.7%)	4(33.3%)	3(25.0%)	3(25.0%)
NISO	98	30(30.6%)	37(37.8%)	11(11.2%)	20(20.4%)
LRD	120	58(48.3%)	15(12.5%)	2(1.7%)	45(37.5%)
ISO	50	23(46.0%)	3(6.0%)	0	24(48.0%)
NISO	70	35(50.0%)	12(17.1%)	2(2.9%)	21(30.0%)
ART	36	20(55.6%)	6(16.7%)	4(11.1%)	6(16.7%)
ISO	12	8(66.7%)	3(25.0%)	0	1(8.3%)
NISO	24	12(50.0%)	3(12.5%)	4(16.7%)	5(20.8%)

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis; TP, true positive; UP, unconfirmed positive; FP, false positive; FN, false negative.

Table 5 The screening efficiency of ultrasound for fetal limb anomalies

	Sensitivity	Specificity
POD	19.1%	99.70%
AHP	76.0%	99.90%
LRD	61.9%	99.90%
Limb	76.0%	100%
Digit	28.9%	100%
Unsp	100%	99.9%
ART	81.3%	99.9%

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis.

during the study, gestational age or indication for examination. The specificity and NPV approached or equaled 100% for each of these abnormalities, reflecting the small number of FP diagnoses.

Pregnancy outcomes are detailed in Table 8. Collectively, the LB rates are reduced, because in large part of the numbers of women electing TOP or experiencing perinatal loss. This is true for both ISO and NISO cases although these conditions in the presence of other malformations exacerbate these trends with even higher rates of adverse outcomes and corresponding lower rates of LB.

The types and frequency of associated malformations are described in Table 9. Each of these conditions is associated with a notable risk for non-limb malformations which is highest for AHP and lowest for POD. Cardiothoracic malformations were the most common associated anomalies in fetuses with AHP and LRD. Facial abnormalities were the most likely in fetuses with POD and ART. For the study population as a whole, cardiothoracic and facial defects were the most common accompanying associated anomalies.

The incidence of aneuploidy in the study population is seen in Table 10. Each of these conditions was associated with an increased risk for aneuploidy, which was as high as 47% for fetuses in the AHP group. Regardless of category, trisomy 18 (T18) was by far the most common chromosome disorder followed by trisomy 13 (T13) and triploidy. The majority of cases of aneuploidy occurred when the limb abnormalities were NISO (86% for LRD and ART and 96% for AHP).

Table 6 The effect of study period, gestational age and indication for examination on ultrasound sensitivity for detection of limb abnormalities

	Study period		Gestational age (weeks)		Indication for examination	
	1990–1999	2000–2009	≤22	>22	Known or suspected malformation	Other
POD	27.7%	15.2%	73.9%	10.0%	35.1%	14.1%
AHP	70.8%	83.3%	79.6%	71.4%	81.8%	65.6%
LRD	62.2%	61.5%	60.3%	65.0%	75.0%	46.3%
ART	66.7%	85.7%	81.0%	75.0%	78.3%	80.0%

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis.

Table 7 The effect of study period, gestational age and indication for examination on the positive predictive values of ultrasound for diagnosis of limb abnormalities

	Study period		Gestational age (weeks)		Indication for examination	
	1990–1999	2000–2009	≤22	>22	Known or suspected malformation	Other
POD	93.3%	91.9%	91.9%	93.3%	96.4%	82.7%
AHP	85.0%	83.3%	82.7%	85.7%	88.5%	80.8%
LRD	97.1%	97.6%	95.9%	100%	98.0%	96.2%
ART	80.0%	85.7%	89.5%	90.0%	94.7%	80.0%

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis.

DISCUSSION

This study presents the accuracy of prenatal diagnosis of common limb abnormalities in a large population referred for a variety of indications. Our findings suggest that overall, AHP, LRD involving the limbs and ART are identified 76% to 81% of the time in a tertiary center with dedicated obstetric sonographers and sonologists. The majority of these abnormalities were NISO. Other than ART, detection was more likely when other major malformations were present. This reflects the fact that one malformation typically prompts a search for others. In our NISO cases, it is unable to be determined which anomaly was detected first, the skeletal or non-skeletal, and prompted the detection of the other. Our data make clear that the finding of POD, AHP, LRD and AHP should prompt a careful search for associated malformations, especially those involving the face and the heart. Our findings also demonstrate that fetuses with these limb abnormalities are at high risk of adverse pregnancy outcome. This reflects the likelihood and severity of associated malformations. Other than ISO cases of POD and LRD, rates of SB, NND and TOP were significant, with correspondingly reduced rates of LB.

Table 8 Pregnancy outcome in cases of fetal limb abnormalities

	N	LB	TOP	STB	NND
POD	330	282(85.5%)	30(9.1%)	4(1.2%)	14(4.2%)
ISO	250	240(96.0%)	7(2.8%)	2(0.8%)	1(0.4%)
NISO	80	42(52.5%)	23(28.8%)	2(2.5%)	13(16.3%)
AHP	110	27(24.5%)	39(35.5%)	22(20.0%)	22(20.0%)
ISO	12	6(50.0%)	3(25.0%)	2(16.7%)	1(8.3%)
NISO	98	21(21.4%)	36(36.7%)	20(20.4%)	21(21.4%)
LRD	120	63(52.5%)	40(33.3%)	9(7.5%)	8(6.7%)
ISO	50	41(82.0%)	8(16.0%)	1(2.0%)	0
NISO	70	22(31.4%)	32(47.7%)	8(11.4%)	8(11.4%)
ART	36	10(27.8%)	12(33.3%)	9(25.0%)	5(13.9%)
ISO	12	6(50.0%)	4(33.3%)	1(8.3%)	1(8.3%)
NISO	24	4(16.7%)	8(33.3%)	8(33.3%)	4(16.7%)

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis; LB, live birth; TOP, termination of pregnancy; STB, stillbirth; NND, neonatal death; ISO, isolated; NISO, non-isolated.

The risk of aneuploidy with all of these disorders warrants consideration of diagnostic testing for aneuploidy and possibly chromosomal microarray analysis depending upon the associated malformations detected. Both PPV and NPV were high, supporting the reliability of ultrasound in demonstrating the presence or absence of these conditions.

Data collected from 17 European registries of congenital malformations (European Surveillance of Congenital Anomalies) for births occurring between 1995 and 1999 revealed 694 cases of LRD. Thirty-six percent were diagnosed prenatally, and 7% were associated with a chromosome anomaly. However, wide variations in the rate of prenatal diagnosis were noted.<sup>4</sup> In a review of prenatal diagnosis of limb reduction deficiencies using data from 20 congenital malformation registries provided by 12 European countries, 250 cases of LRD were identified. Of these, 55% were ISO, and 45% had associated malformations. The prenatal detection rate of ISO cases was 24.6% versus 49.1% of NISO cases. However, associated anomalies were not specified, and there was significant geographic variation with detection rates ranging from 20% to 64%. The TOP rate was 9.4% for ISO cases and 25.2% for those with associated malformations.<sup>5</sup>

Kevern and colleagues reviewed two study populations to examine the accuracy of prenatal sonographic identification of fetal limb abnormalities. In a retrospective review of 60 infants referred for genetic counseling, for the birth of a child with a limb abnormality, 15 were detected prenatally for a sensitivity of 25%. Using the Wessex Antenatally Detected Anomalies Register, 67 cases of limb anomaly were diagnosed prenatally, 56 of which were confirmed postnatally (PPV 84%). Correctly diagnosed abnormalities included micromelia, rhizomelia, absent bone, shortened bone and POD. Only 3/56 had associated malformations (5.4%).<sup>6</sup>

A retrospective review of 64 fetuses with a short, absent or abnormal radius and/or ulna revealed 15% were ISO, 70% were correctly diagnosed prenatally and 84% were confirmed postnatally. Chromosomal anomalies and genetic syndromes occurred in 29.7% and were more likely if the abnormalities were bilateral.<sup>7</sup>

The Northern Congenital Abnormalities Survey collects data on all major congenital abnormalities in the northern region of England. During the period 2000 to 2005, there were 18 cases of radial ray defects. Eleven (61%) were diagnosed prenatally. TOP was elected in 12 cases, which included four fetuses with T18 and three others with a non-chromosomal syndrome. Three-dimensional (3D) ultrasound was used in 8/18 cases and either confirmed the ultrasound findings or allowed the diagnosis of additional anomalies. It was felt to be especially useful in illustrating the relationships of the fetal forearm and hand and for examination of the facial anatomy.<sup>8</sup>

A prior study from this institution included 100 cases of upper extremity anomalies evaluated in the congenital hand surgery clinic. These children were not necessarily a part of the database for the current study and most had been imaged prenatally at other sites. Thirty-one of these cases had an upper extremity abnormality identified on prenatal ultrasound. In 18/31 (58%), the prenatal diagnosis was confirmed postnatally.<sup>9</sup>

In a review of 271 cases of LRD occurring in 11 maternity hospitals in northeastern France during a 26-year period



Table 9 Non-orthopedic malformations associated with fetal limb abnormalities

	N	CNS	Face	Nuchal	Cardiothoracic	Abdom	Renal
POD	330	13 (3.9%)	32 (9.7%)	9 (2.7%)	27 (8.2%)	15 (4.5%)	17 (5.2%)
AHP	110	41 (37.3%)	50 (45.5%)	19 (17.3%)	65 (59.1%)	36 (32.7%)	36 (32.7%)
LRD	120	22 (18.3%)	18 (15.0%)	5 (4.2%)	29 (24.2%)	22 (18.3%)	24 (20.0%)
ART	36	8 (22.2%)	12 (33.3%)	5 (13.9%)	9 (25.0%)	7 (19.4%)	5 (13.9%)
	596	84 (14.1%)	112 (18.8%)	38 (6.4%)	130 (21.8%)	80 (13.4%)	82 (13.8%)

CNS, central nervous system; POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis.

(1979–2004), the overall birth prevalence was 7.8/10 000. The LRD were ISO in 114 (42.1%) and associated with malformations in 157 (57.9%). Chromosomal abnormalities were identified in 17 (6.3%). From the beginning to the end of the study period, the prenatal detection rates ranged from 4.0% to 29.5% for ISO cases and 18.2% to 56.2% for NISO cases. Corresponding rates of TOP were 11.5% to 29.3%. The most common associated malformations in this population involved the urogenital system (14.2%), cardiovascular system (11.4%) and central nervous system (7.7%).<sup>10</sup>

Our detection rates meet or exceed those reported in other series. It should be appreciated that although most cases of the functionally significant conditions of LRD of the long bones and ART can be reliably detected by routine imaging of the arms and legs, 20% to 25% may still be missed. Given the experience of the personnel involved, the detection rates for the conditions described are thought to represent a near maximum of what could reasonably be expected following current guidelines. Lower levels of detection would be expected in smaller centers with less volume and expertise in fetal imaging. This is also suggested by the wide regional variations described in multicenter reports. Extra or missing digits are more likely to escape detection, which is not surprising because imaging of the hands and feet is not stipulated for the screening obstetric ultrasound. Our findings demonstrate the feasibility and advisability of including examination of the hands and digits when major malformations are identified and vice versa. Abnormal posturing of the hands was readily appreciated, especially in conjunction with other malformations. This information can be useful in refining the prognosis when discussing the findings with the patient. Our experience also makes clear that imaging the arms and legs provides an excellent opportunity to rule out abnormalities

of the joints in addition to malformations and deformations of the extremities. The failure to monitor fetal movement has resulted in more than 75% of cases remaining undetected until birth.<sup>11,12</sup> In our population, over 90% of cases of ISO ART were detected prenatally. The high likelihood of physical and/or mental impairment, perinatal loss and TOP with this condition suggests movement of the joints merits attention when documenting the presence and morphology of the extremities.

Detection of AHP and ART improved from the first half of the study period to the second. This may reflect improvement in technology as well as a greater appreciation of the significance of these conditions, prompting more detailed imaging. All conditions were more likely to be diagnosed when the index of suspicion was elevated. Diagnosis of these conditions, other than POD, was not significantly affected by gestational age, suggesting they are capable of being identified regardless of fetal size.

The strengths of our study include a large population referred for a variety of indications to a single center with a standard approach to sonographic examination of the fetal anatomy. The number and type of limb abnormalities is greater than reported from other single centers. All TP, FP, true negative and FN diagnoses were confirmed by postnatal findings. The limitations include the fact that neonatal follow-up is available for 90% to 95% of our population, not 100%. Because neonatal confirmation of prenatal ultrasound findings was unavailable in our UP cases, it is possible that our detection rates may have been overestimated and our FN rates underestimated. Because our study covers a 20-year period, it is not possible to accurately account for the effect of changes in technology. Our ultrasound equipment has been serially replaced over this time and we are

Table 10 Aneuploidy incidence and type associated with fetal limb abnormalities

	N	T18	T13	T21	Triploidy	Other
POD	12/330 (3.6%)	10 (83.3%)	1 (8.3%)		1 (8.3%)	
AHP	52/110 (47.3%)	38 (73.1%)	5 (9.6%)	1 (1.9%)	2 (3.8%)	6 (11.5%)
LRD	6/120 (5.0%)	4 (66.6%)				2 (33.3%)
ART	7/36 (19.4%)	5 (71.4%)			1 (14.3%)	1 (14.3%)

POD, polydactyly; AHP, abnormal hand position; LRD, limb reduction defect; ART, arthrogryposis.

unable to identify which examinations were performed on which machines. 3D ultrasound was employed in some of the later studies, but its contribution to the diagnosis of the limb abnormalities or associated malformations cannot reliably be determined from this data set.

## CONCLUSION

In summary, imaging of the fetal limbs can allow the accurate diagnosis of LRD of the long bones, ART and, if looked for, AHP in 76% to 81% of cases overall. POD is less often diagnosed. All of these anomalies, other than ISO POD, are associated with increased risk for associated non-skeletal malformations, aneuploidy, STB, NND and pregnancy termination. This information can be useful in directing evaluation and management, counseling patients and guiding delivery planning.

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## WHAT'S ALREADY KNOWN ABOUT THIS TOPIC?

- Current guidelines recommend documentation of the arms and legs as a component of the standard fetal ultrasound examination.
- There is a wide regional variation in the types and ultrasound detection rates of fetal limb abnormalities using prenatal ultrasound.

## WHAT DOES THIS STUDY ADD?

- The accuracy of ultrasound diagnosis of major and minor limb abnormalities in a single center with a uniform approach to fetal imaging.
- It is advisable and feasible to examine the hands and digits when non-skeletal malformations are identified.
- Attention to limb and joint mobility, in addition to limb presence and morphology, allows the prenatal detection of many cases of arthrogryposis.