

**American College of Radiology
ACR Appropriateness Criteria®
Nuchal Translucency Evaluation at 11 to 14 Weeks Gestational Age**

Variant: 1 Routine nuchal translucency measurement at 11 to 14 weeks of gestation for single or twin gestations. Initial imaging.

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal	Usually Appropriate	0
US pregnant uterus transvaginal	May Be Appropriate	0
US duplex Doppler pregnant uterus	Usually Not Appropriate	0
US echocardiography fetal	Usually Not Appropriate	0

Variant: 2 Increased nuchal translucency measurement at 11 to 14 weeks of gestation for single gestation.

Procedure	Appropriateness Category	Relative Radiation Level
US echocardiography fetal	Usually Appropriate	0
US pregnant uterus transabdominal	Usually Appropriate	0
US duplex Doppler pregnant uterus	May Be Appropriate	0
US pregnant uterus transvaginal	May Be Appropriate	0

Variant: 3 Increased nuchal translucency in dichorionic twins at 11 to 14 weeks of gestation.

Procedure	Appropriateness Category	Relative Radiation Level
US echocardiography fetal	Usually Appropriate	0
US pregnant uterus transabdominal	Usually Appropriate	0
US duplex Doppler pregnant uterus	May Be Appropriate	0
US pregnant uterus transvaginal	May Be Appropriate	0

Variant: 4 Increased nuchal translucency in monochorionic twins at 11 to 14 weeks of gestation.

Procedure	Appropriateness Category	Relative Radiation Level
US echocardiography fetal	Usually Appropriate	0
US pregnant uterus transabdominal	Usually Appropriate	0
US duplex Doppler pregnant uterus	May Be Appropriate	0
US pregnant uterus transvaginal	May Be Appropriate	0

Panel Members

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Summary of Literature Review

Introduction/Background

Singletons

Nuchal translucency is defined as the hypoechoic space between the overlying skin and underlying soft tissues of the posterior cervical spine of the first trimester fetus. Below a defined threshold, this translucent space at the back of the fetal neck is a normal ultrasound (US) finding at 11 to 14 weeks of gestation; above this threshold, the fetus is considered to have an increased nuchal translucency, which is a marker for fetal aneuploidy, genetic syndromes, structural anomalies, and intrauterine demise [1]. Fetal nuchal translucency increases with crown-rump length, so gestational age must be taken into account when determining whether a given nuchal translucency thickness is increased. It is customary to quantify the deviation of the nuchal translucency measurement from normal using percentiles or multiples of the median; an alternate approach is to use an absolute threshold value of ≥ 3 mm, which is uniformly increased at any crown-rump length between 11 and 14 weeks [2]. As a marker for adverse fetal outcomes, these risks increase as the nuchal translucency thickness increases.

Nuchal translucency is a powerful and independent US marker for fetal aneuploidy. Since the first report of transient nuchal fluid in an 11 week fetus with Down syndrome nearly 30 years ago, numerous studies have confirmed a strong association between increased nuchal translucency and abnormal fetal karyotype [1,3-5]. In an early prospective screening study, 6% of fetuses were found to have a nuchal translucency of ≥ 3 mm at 11 to 14 weeks of gestation with a 10-fold increase in chromosomal abnormalities such as trisomies, Turner syndrome, and triploidy [5]. Follow-up studies confirmed that fetal nuchal translucency combined with maternal age could detect over 75% of cases of trisomy 21 with a false positive rate of about 5% [6,7]. The American College of Obstetricians and Gynecologists (ACOG) recommends offering aneuploidy screening to all patients, acknowledging the advantages of early screening [2]. The most common first trimester screen generates individualized risk estimates based on combining the size of the nuchal translucency according to crown-rump length, levels of maternal serum free beta subunit of human chorionic gonadotropin and pregnancy-associated plasma protein A and the maternal age-adjusted background risk for aneuploidy, with or without nasal bone assessment. A nuchal translucency measurement of ≥ 3 mm places the fetus at sufficient risk with minimal benefit in waiting for combined risk calculations irrespective of gestational age [8].

Twins

As an independent marker of fetal aneuploidy, nuchal translucency is particularly useful in twins because of the inability of maternal serum markers or age to identify the fetus at risk. Nuchal translucency measurement allows each fetus of a twin pregnancy to be assessed individually; the distribution of measurements does not differ significantly between singletons and twins so that standard thresholds can be used [9,10]. In dichorionic twins, fetus-specific risks are calculated on the assumption each twin has an independent risk reflected by its own nuchal translucency [2,11,12]. In monochorionic twins, a pregnancy-specific risk is calculated using the average nuchal translucency thickness of both fetuses [2,13]. As in singletons, increased nuchal translucency may also be a marker of fetal structural anomalies, genetic syndromes, and intrauterine demise in twins.

Special Imaging Considerations

Measurement of the nuchal translucency has been standardized in order to improve its performance in screening for fetal aneuploidy. Quality assurance programs for nuchal translucency assessment have been established by the Fetal Medicine Foundation in London and the Nuchal Translucency Quality Review in the United States. The ACR Appropriateness Criteria methodology

assumes that each imaging procedure is performed and interpreted by an expert, but it should be noted that there remains considerable inter- and intra-observer variability in nuchal translucency measurements, highlighting the importance of ongoing quality assessment [14,15].

First trimester screening algorithms can be improved by assessing for the presence or absence of the nasal bone and adjusting aneuploidy risk calculations accordingly [16]. The absence of the nasal bone at 11 to 14 weeks is another powerful US marker of fetal aneuploidy; it improves the performance of early screening by decreasing the false-positive rate to 2.5% [16]. Other first trimester US markers of fetal aneuploidy such as increased frontomaxillary facial angle, an aberrant right subclavian artery, presence of tricuspid regurgitation, reversed a-wave in the ductus venosus, and increased iliac wing angle have not been incorporated into routine screening algorithms and their clinical usefulness in the general population remains uncertain [17]. The role of nuchal translucency measurement in the new era of cell-free fetal DNA screening is also uncertain. It is recommended that a first trimester US for the sole purpose of nuchal translucency screening not be performed in patients with negative cell-free fetal DNA [18].

Initial Imaging Definition

Initial imaging is defined as imaging at the beginning of the care episode for the medical condition defined by the variant. More than one procedure can be considered usually appropriate in the initial imaging evaluation when:

- There are procedures that are equivalent alternatives (ie, only one procedure will be ordered to provide the clinical information to effectively manage the patient's care)

OR

- There are complementary procedures (ie, more than one procedure is ordered as a set or simultaneously wherein each procedure provides unique clinical information to effectively manage the patient's care).

Discussion of Procedures by Variant

Variant 1: Routine nuchal translucency measurement at 11 to 14 weeks of gestation for single or twin gestations. Initial imaging.

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A. US duplex Doppler pregnant uterus

Although the utility of Doppler imaging has been investigated in research protocols, there is no literature to support a recommendation to universally incorporate Doppler studies into routine first trimester screening algorithms for fetal aneuploidy [17]. Given the theoretical risk of thermal damage to the developing fetus from the use of color and pulsed Doppler US, its use is not advised when the nuchal translucency is of normal thickness or below the threshold of 3 mm at 11 to 14 weeks of gestation [2,19].

Variant 1: Routine nuchal translucency measurement at 11 to 14 weeks of gestation for single or twin gestations. Initial imaging.

B. US echocardiography fetal

In the absence of other maternal or fetal risk factors, there is no literature to support the routine use of fetal echocardiography in patients with normal nuchal translucency measurements at 11 to 14 weeks of gestation [20].

Variant 1: Routine nuchal translucency measurement at 11 to 14 weeks of gestation for single or twin gestations. Initial imaging.

C. US pregnant uterus transabdominal

ACOG recommends aneuploidy screening or diagnostic testing be offered to all women in early pregnancy [2]. For women who choose screening over invasive diagnostic testing, first trimester combined screening with nuchal translucency and maternal serum markers remains a reasonable option for single and twin gestations [2,18,21]. Although ACOG does not endorse a specific screening test, citing that no one screening test is superior to another in all test characteristics, there are several benefits of first trimester screening, including maternal privacy, early reassurance, and timely detection of fetal abnormalities with the option for earlier and safer pregnancy termination [2]. Using the nuchal translucency measurement and maternal serum pregnancy-associated plasma protein A and free beta subunit of human chorionic gonadotropin levels, most laboratories will report specific risk estimates for trisomy 21, 18, and 13 after adjusting for crown-rump length and maternal factors such as age, prior history of aneuploidy, weight, and race [2]. In clinical practice, first trimester combined screening for aneuploidy detects about 90% of affected pregnancies at a false positive rate of 5% for single gestations with slightly lower sensitivities reported for twins [12,17].

The nuchal translucency measurement can be obtained using transabdominal US in about 95% of patients [5,17,22,23]. An increased nuchal translucency is defined as a measurement ≥ 3 mm or above the 99th percentile for the crown-rump length [2]. Although a normal nuchal translucency is reassuring, nuchal translucency assessment should not be used in isolation to determine risk because of its inferior performance, detecting only 70% of trisomy 21 fetuses [17,24]. The nuchal US should also not be performed as a screening test for fetal anomalies in low-risk women or replace the standard second trimester anatomic fetal survey. Although there is a recognized association between increased nuchal translucency and fetal structural malformations, it is estimated that only 50% of major fetal anomalies can be detected prior to 14 weeks with a false positive rate of 3% to 4% at this early gestational age [25,26]. Despite a strong association between increased nuchal translucency and major cardiac anomalies, nuchal translucency is also not a good screening test for congenital heart disease. In a large meta-analysis, <25% of major heart defects were detected using a nuchal translucency cut-off at the 99th percentile in euploid fetuses, confirming that most cases of congenital heart disease have normal nuchal translucency thickness [27].

Variant 1: Routine nuchal translucency measurement at 11 to 14 weeks of gestation for single or twin gestations. Initial imaging.

D. US pregnant uterus transvaginal

Transvaginal US may be utilized for first trimester aneuploidy screening if optimal views of the nuchal translucency cannot be obtained using the transabdominal approach, which occurs in about 5% of patients [5,24,28]. The higher resolution of transvaginal US can be particularly helpful when imaging women with high body mass indexes. The transvaginal approach may also be favored in patients with multiple abdominal surgeries or presenting with a transverse fetal lie in the lower aspect of the uterus. However, reduced probe mobility limits the available image planes, so using a combination of transabdominal and transvaginal US may be necessary to fully assess the first

trimester fetus. Fetal nuchal translucency measurements are similar whether measured by transabdominal or transvaginal US as long as the same principles for image acquisition and nuchal translucency measurement are applied [29].

The transvaginal approach may produce better quality images to assess the nasal bone than transabdominal US [65]. However, perhaps because of the restricted range of movement, a 3-fold higher failure of nasal bone imaging has been reported with transvaginal US [30]. If there is difficulty obtaining the nuchal translucency measurement, there is likely to be difficulty in obtaining the proper image to assess for the presence or absence of nasal bone regardless of approach.

Variant 2: Increased nuchal translucency measurement at 11 to 14 weeks of gestation for single gestation.

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A. US duplex Doppler pregnant uterus

Aneuploidy risk can be further investigated by assessing flow across the tricuspid valve or in the ductus venosus; in cases of increased nuchal translucency, the probability of fetal aneuploidy is higher when significant tricuspid regurgitation or reversed a-waves in the ductus venosus are present [31]. Although these markers have not been incorporated into routine clinical practice, their evaluation may improve the performance of first trimester aneuploidy screening or assist with patient counseling when the nuchal translucency is ≥ 3 mm [17,31].

Normal antegrade flow across the tricuspid valve can be evaluated by both color and pulsed Doppler. Although a small amount of tricuspid regurgitation is not uncommon in the first trimester, retrograde flow beyond a third of the right atrial diameter on color Doppler or with a duration $\geq 50\%$ of ventricular systole on pulsed Doppler interrogation is considered significant [32]. Tricuspid regurgitation is observed in 1% of euploid fetuses compared to over half of fetuses with trisomy 21 and about a third of fetuses with trisomy 18, 13, or Turner syndrome [32]. A recent review reported that first trimester tricuspid regurgitation was associated with a 10-fold increase in congenital heart disease [33].

The ductus venosus can be easily identified by its accelerated flow velocity on color Doppler and then interrogated with pulsed Doppler to assess its flow characteristics. The triphasic waveform of the ductus venosus, reflecting the phases of the cardiac cycle, normally has forward flow toward the heart at all times; a reversed a-wave, corresponding to retrograde flow during atrial contraction, is abnormal. Although the cause of this abnormality in fetal aneuploidy is uncertain, a reversed a-wave is seen in 3% to 4% of euploid fetuses compared to about 70% of trisomy 21 and trisomy 18, 65% of trisomy 13, and 75% of Turner syndrome fetuses [34]. The addition of ductus venosus Doppler assessment to nuchal translucency measurement improves the detection rate of trisomy 21 to 96% with 3% false positive rate [34]. Ductus venosus a-wave abnormalities are also associated with congenital heart disease [35]. In chromosomally normal fetuses with an increased nuchal translucency, a reversed a-wave in the ductus venosus is associated with a 3-fold increase in the likelihood of a major heart anomaly; a normal ductus venosus waveform decreases the risk of congenital heart disease by 50% [36]. The addition of Doppler studies mandates application of the as low as reasonably achievable principle to minimize fetal risk [19]. Although first trimester tricuspid regurgitation and ductus venosus reversed a-wave increase the likelihood of fetal

aneuploidy and congenital heart defects, Doppler studies should be reserved to fetuses at risk such as those with a nuchal translucency ≥ 3 mm [37].

Variant 2: Increased nuchal translucency measurement at 11 to 14 weeks of gestation for single gestation.

B. US echocardiography fetal

Although nuchal translucency does not perform well as a screening test for congenital heart disease, a nuchal translucency at ≥ 3 mm or ≥ 99 th percentile is an accepted indication for fetal echocardiography [2,20,38,39]. The optimal time to assess the fetal heart is at 18 to 22 weeks of gestation; however, early echocardiography may be performed [20]. One of the major benefits of first trimester fetal echocardiography is early reassurance in at-risk pregnancies. There are also advantages of early detection of congenital heart disease including timely investigation for associated chromosomal or genetic conditions, extra-cardiac anomalies, and the option for earlier and safer pregnancy termination. A complete cardiac evaluation is possible at 13 to 14 weeks in over 90% of cases [40]. Even without a full evaluation, first trimester fetal echocardiography is possible at 11 to 14 weeks of gestation with a high degree of accuracy, identifying 50% to 65% of major heart anomalies in low- and high-risk patients, respectively [26]. At this early gestational age, about half of hypoplastic left heart and atrioventricular septal defects can be detected compared to less than a quarter of conotruncal abnormalities [41]. The detection rate of congenital heart disease in the first trimester varies according to the experience of the center, the population studied, and the type of defect present; first trimester pitfalls can result in both false positive and missed diagnoses which necessitates caution [42].

Because of the limitations of early cardiac imaging, a repeat fetal echocardiogram is still recommended in the second trimester in pregnancies with increased nuchal translucency [20,43]. At a nuchal translucency threshold of ≥ 3 mm or ≥ 99 th percentile, about 1% of patients undergoing first trimester nuchal translucency screening will require standard second trimester fetal echocardiography [43]. Given the small number of referrals and the high prevalence of major cardiac defects in this group, the performance of second trimester fetal echocardiography to detect congenital heart disease in cases of increased nuchal translucency should be close to 80% in echocardiography units [44].

Variant 2: Increased nuchal translucency measurement at 11 to 14 weeks of gestation for single gestation.

C. US pregnant uterus transabdominal

During first trimester aneuploidy screening, a nuchal translucency measurement of ≥ 3 mm places the fetus at sufficient risk that genetic counseling and invasive diagnostic testing should be promptly offered [8]. Studies have shown that there is minimal benefit in waiting for maternal serum results or calculated risks from the laboratory; about one-third of fetuses with nuchal translucency thickness above this threshold will have a chromosomal abnormality and half of these will be trisomy 21 [8,45,46]. Although immediate chorionic villus sampling shortens the time to obtain a definitive diagnosis, offering secondary screening with cell-free fetal DNA from maternal blood is also supported by ACOG for women who initially screen positive on first trimester combined screening or have a nuchal translucency ≥ 3 mm [2,21]. However, cell-free fetal DNA screening only detects trisomy 21, 18, and 13 and sex chromosome aneuploidies, so failure to undergo invasive diagnostic testing will miss certain genetic causes of increased nuchal translucency such as Noonan syndrome and 22q11.2 deletion syndrome [2]. In euploid fetuses with nuchal translucencies ≥ 3 mm, 10% have genetic variants consistent with Noonan syndrome;

targeted genetic studies including microarray analysis are now recommended in all cases of increased nuchal translucency [47,48].

When the nuchal translucency measures ≥ 3 mm, the finding of additional first trimester US markers of fetal aneuploidy, such as absence of the nasal bone and increased frontomaxillary and iliac angles, may be useful for patient counseling but the ≥ 3 mm thickness with or without additional markers warrants further evaluation [17,31,45,49]. In experienced centers, an early fetal anatomic survey may also be performed at the time an increased nuchal translucency is identified because of the risk of major structural anomalies, even in fetuses subsequently found to be euploid. This provides the opportunity for the early detection of major malformations affecting the brain, heart, abdominal wall, and limbs [26]. However, the sensitivity of an US at 11 to 14 weeks for the detection of congenital anomalies is only about 50% compared to 75% at 18 to 22 weeks [50]. Although fetal anomalies and congenital heart disease are more common in cases of aneuploidy, these risks remain elevated in euploid fetuses with a history of a nuchal translucency ≥ 3 mm or a first trimester cystic hygroma [22,24]. Therefore, these patients who are continuing their pregnancies should be offered a second trimester anatomic survey and a fetal echocardiogram irrespective of the fetal DNA analysis [2,19,20].

The risk of an adverse pregnancy outcome is proportional to the degree of nuchal translucency enlargement; there is an increased risk of intrauterine demise in fetuses with large nuchal translucencies, even in the absence of associated chromosomal or structural abnormalities [22,23]. Although there are no studies that specifically address the optimal management of these pregnancies after the initial US evaluation and diagnostic testing, serial fetal surveillance is usually added to routine prenatal care with periodic US examinations to monitor fetal growth and well-being [2,45].

Variant 2: Increased nuchal translucency measurement at 11 to 14 weeks of gestation for single gestation.

D. US pregnant uterus transvaginal

A transvaginal US may be advantageous in patients with an increased nuchal translucency, particularly if planning to assess for additional first trimester markers of fetal aneuploidy or screen for major anomalies in women with increased body mass indexes, abdominal wall scarring, or fetal positioning in the lower uterus. Fetal nuchal translucency measurements are similar as long as the same principles for image acquisition are applied, so obtaining a transvaginal measurement is not required if transabdominal imaging is satisfactory [29]. However, high resolution transvaginal US may more clearly identify the amnion and nuchal translucency borders to ensure an accurate maximal measurement in patients with suboptimal transabdominal views.

Variant 3: Increased nuchal translucency in dichorionic twins at 11 to 14 weeks of gestation.

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A. US duplex Doppler pregnant uterus

Although there is limited published data on the use of color and pulsed Doppler in screening twin pregnancies, both the presence of significant tricuspid regurgitation and reversed a-waves in the ductus venosus waveform are associated with chromosomal and cardiac defects in dichorionic twins with increased nuchal translucency [51]. Although these specialized studies are not typically performed in patients with uncomplicated dichorionic twins, Doppler imaging is reasonable to further evaluate risk in cases of nuchal translucency ≥ 3 mm affecting one or both fetuses [51]. The

role of Doppler US in the follow-up surveillance of dichorionic twins is uncertain in the absence of intrauterine growth restriction [52].

Variante 3: Increased nuchal translucency in dichorionic twins at 11 to 14 weeks of gestation.

B. US echocardiography fetal

An increased nuchal translucency in a dichorionic twin is an indication to obtain a fetal echocardiogram [20]. Performing fetal echocardiography on twins is challenging because of both maternal factors, such as increased body mass index or inability to rest comfortably during a lengthy examination, and fetal factors, such as unfavorable fetal position or limited views due to the presence of a co-twin. Although early fetal echocardiography may be attempted, a standard fetal echocardiogram at 18 to 22 weeks with an expected detection rate of over 80% for major cardiac anomalies is recommended when increased nuchal translucency is detected in dichorionic twins [26,44].

Variante 3: Increased nuchal translucency in dichorionic twins at 11 to 14 weeks of gestation.

C. US pregnant uterus transabdominal

Although no screening method in twins is as accurate as it is in singletons, first trimester combined screening in dichorionic twins provides detection rates of fetal aneuploidy close to that reported in singletons with sensitivities over 85% [53,54]. In the majority of twin pregnancies, acceptable images of the nuchal translucency can be acquired by transabdominal US using the same principles to measure its thickness. As in singleton pregnancies, a threshold ≥ 3 mm at 11 to 14 weeks of gestation is used to define increased nuchal translucency, and it has the same implications in dichorionic twins as it does in singletons as a marker for aneuploidy, genetic syndromes, structural anomalies, and intrauterine death [2]. The mode of conception and use of various assisted reproductive techniques does not appear to have a significant impact on the frequency of increased nuchal translucency measurements [55].

One of the advantages of nuchal translucency screening in dichorionic twins is the ability to perform individual measurements on each fetus and generate fetus-specific risks [45]. The management of a positive screen or an increased nuchal translucency is similar in twins and singletons; genetic counseling, diagnostic testing, and in continuing pregnancies with a history of increased nuchal translucency or diagnosed with fetal aneuploidy, a detailed anatomic survey, and fetal echocardiography in the second trimester are recommended. The prevalence of increased nuchal translucency in dichorionic twins with a normal karyotype is similar to that in singletons [56]. Although an individual risk can be estimated for each twin and a patient may opt for only sampling the twin suspected to be at risk, it is customary to sample both twins at the time of diagnostic testing to avoid a missed diagnosis because of averaged maternal biochemistries or incorrect assignment of chorionicity [57]. For those who decline diagnostic testing, an evaluation for additional first trimester markers of fetal aneuploidy such as the presence or absence of the nasal bone may assist in patient counseling; however, nasal bone assessment is more challenging in twin pregnancies [58].

After the second trimester anatomy survey and fetal echocardiogram, there are no robust evidence-based recommendations for the US follow-up of dichorionic twins with increased nuchal translucency. However, given the risk for intrauterine demise, it is reasonable that serial surveillance of fetal growth and well-being be performed in the third trimester [45,59,60].

Variante 3: Increased nuchal translucency in dichorionic twins at 11 to 14 weeks of gestation.

D. US pregnant uterus transvaginal

As in singleton pregnancies, transvaginal US can be utilized at 11 to 14 weeks of gestation to assess the nuchal translucency, additional markers of aneuploidy, as well as early fetal anatomy in dichorionic twins. Fetal nuchal translucency measurements are similar as long as the same principles for image acquisition and measurement are employed [29]. Given the larger uterine dimensions from the presence of two fetuses, two sacs of amniotic fluid, and two placentas, a combination of both transabdominal and transvaginal imaging may be optimal to fully assess dichorionic twins with increased nuchal translucency.

Variant 4: Increased nuchal translucency in monochorionic twins at 11 to 14 weeks of gestation.

Variant 4: Increased nuchal translucency in monochorionic twins at 11 to 14 weeks of gestation.

A. US duplex Doppler pregnant uterus

The detection of abnormal flow across the tricuspid valve or in the ductus venosus by color and pulsed Doppler increases the likelihood of fetal aneuploidy in monochorionic twins with increased nuchal translucency. In addition, both reversed or absent a-waves in the ductus venosus have been reported to be markers for congenital heart disease and the development of twin-twin transfusion syndrome later in gestation [61,62]. Although Doppler imaging is likely of little to no value in uncomplicated monochorionic twins, serial assessment of the ductus venosus, umbilical artery and vein, and middle cerebral artery may be useful in cases complicated by unequal placental sharing, twin-twin transfusion syndrome, or twin anemia polycythemia sequence [45].

Variant 4: Increased nuchal translucency in monochorionic twins at 11 to 14 weeks of gestation.

B. US echocardiography fetal

Because of the substantial risk of congenital heart disease, there is strong evidence to support the use of fetal echocardiography for all monochorionic twins, regardless of the nuchal translucency measurement [20]. The overall risk of congenital heart disease in monochorionic twins is 2%, double the background risk of a singleton, and increases to 5% in cases of twin-twin transfusion syndrome, particularly among recipient twins [63]. Recipient twins often demonstrate evidence of volume overload with increased pulmonary and aortic velocities, cardiomegaly, and atrioventricular regurgitation, which, over time, can result in biventricular hypertrophy and diastolic dysfunction [64]. Poor right ventricular systolic function can lead to functional right ventricular outflow tract obstruction in up to 10% of recipient twins, which may progress to severe pulmonic stenosis and a poor prognosis [64]. In contrast, recipient twins with normal cardiac function have improved survival [65]. Therefore, in addition to excluding structural heart defects in monochorionic twins, fetal echocardiography with a detailed functional assessment may be useful in identifying cases of twin-twin transfusion syndrome that would benefit from fetoscopic laser therapy and in evaluating the response to treatment. Although an increased nuchal translucency may prompt early echocardiography in monochorionic twins, standard second trimester fetal echocardiography is warranted in all monochorionic twins because of the substantial risk of both structural and functional heart abnormalities regardless of the nuchal translucency measurements [59,65].

Variant 4: Increased nuchal translucency in monochorionic twins at 11 to 14 weeks of gestation.

C. US pregnant uterus transabdominal

First trimester combined screening with nuchal translucency and maternal serum markers is

currently a common choice for aneuploidy screening in monochorionic twins [53,54]. In monochorionic twins, each fetus is assumed to have the same risk of aneuploidy equivalent to the maternal age risk of a singleton. Therefore, aneuploidy risk estimates for trisomy 21, 18, and 13 are calculated using the mean nuchal translucency measurement of the twins, usually obtained via transabdominal US. Overall, first trimester combined screening in monochorionic twins provides detection rates of fetal aneuploidy similar to that reported in singletons but with a higher false positive rate [11,53]. A meta-analysis of first trimester combined aneuploidy screening reported a sensitivity of 87.4% for monochorionic twins compared to 86.2% for dichorionic twins [54]. Nuchal translucency measurements have been observed to be higher in monochorionic twins compared to dichorionic twins, which may explain the higher false-positive rate of aneuploidy screening in monochorionic twin pregnancies. In monochorionic twins with increased nuchal translucency thickness, an assessment for additional first trimester markers of fetal aneuploidy, such as the absence of the nasal bone, may be helpful to patients trying to decide whether to undergo invasive diagnostic testing [58]. Increased nuchal translucency thickness in a monochorionic twin may also be an early manifestation of inter-fetal transfusion and early hypervolemic congestion in a recipient twin. Indeed, both nuchal translucency abnormalities and crown-rump length discrepancies have been associated with an increased risk of twin-twin transfusion syndrome [66-68]. A crown-rump discrepancy of >10% is associated with almost a doubling the risk of developing twin-twin transfusion syndrome [67,68]. Nuchal translucency discordance >20% has been associated with a >30% risk of severe twin-twin transfusion syndrome and early fetal death compared with a <10% risk when the discordance is <20% [69].

As in singletons and dichorionic twins, an increased nuchal translucency in monochorionic twins is associated with fetal aneuploidy, genetic syndromes, structural anomalies, and intrauterine demise. Genetic counseling, invasive diagnostic testing, fetal anatomic surveys, and fetal echocardiography are recommended in monochorionic twin pregnancies complicated by increased nuchal translucency thickness affecting one or both twins. Serial US surveillance of monochorionic twins is advised because of complications associated with sharing a single placenta such as twin-twin transfusion syndrome, unequal placental sharing with discordant twin growth and selective intrauterine growth restriction, and twin anemia polycythemia sequence [64,70]. Because of these unique complications, women with monochorionic pregnancies are followed more closely than dichorionic pregnancies; serial US evaluations every 2 weeks starting at 16 weeks until delivery should be considered [45]. Despite its potential value, a recent review concluded that it is not currently possible to predict adverse outcomes in monochorionic twin pregnancies based on nuchal translucency assessment alone [71].

Variant 4: Increased nuchal translucency in monochorionic twins at 11 to 14 weeks of gestation.

D. US pregnant uterus transvaginal

Transvaginal US at 11 to 14 weeks may be utilized to assess nuchal translucency, additional first trimester markers of aneuploidy, early anatomy, and cardiac structure in monochorionic twins when acceptable images cannot be obtained transabdominally. Fetal nuchal translucency measurements are comparable between transabdominal and transvaginal US as long as the same principles for image acquisition and measurement are followed [29].

Summary of Recommendations

- **Variation 1:** US pregnant uterus transabdominal is usually appropriate for the initial imaging of routine nuchal translucency measurement at 11 to 14 weeks of gestation for single or twin gestations.
- **Variation 2:** US echocardiography fetal and US pregnant uterus transabdominal are usually appropriate for the evaluation of increased nuchal translucency measurement at 11 to 14 weeks of gestation for single gestation. These procedures are complementary (ie, more than one procedure is ordered as a set or simultaneously in which each procedure provides unique clinical information to effectively manage the patient's care).
- **Variation 3:** US echocardiography fetal and US pregnant uterus transabdominal are usually appropriate for the evaluation of increased nuchal translucency in dichorionic twins at 11 to 14 weeks of gestation. These procedures are complementary (ie, more than one procedure is ordered as a set or simultaneously in which each procedure provides unique clinical information to effectively manage the patient's care).
- **Variation 4:** US echocardiography fetal and US pregnant uterus transabdominal are usually appropriate for the evaluation of increased nuchal translucency in monochorionic twins at 11 to 14 weeks of gestation. These procedures are complementary (ie, more than one procedure is ordered as a set or simultaneously in which each procedure provides unique clinical information to effectively manage the patient's care).

Safety Considerations in Pregnant Patients

Imaging of the pregnant patient can be challenging, particularly with respect to minimizing radiation exposure and risk. For further information and guidance, see the following ACR documents:

- ACR-SPR Practice Parameter for the Safe and Optimal Performance of Fetal Magnetic Resonance Imaging (MRI)
- ACR-SPR Practice Parameter for Imaging Pregnant or Potentially Pregnant Patients with Ionizing Radiation
- ACR-ACOG-AIUM-SMFM-SRU Practice Parameter for the Performance of Standard Diagnostic Obstetrical Ultrasound
- ACR Manual on Contrast Media
- ACR Manual on MR Safety

Supporting Documents

The evidence table, literature search, and appendix for this topic are available at <https://acsearch.acr.org/list>. The appendix includes the strength of evidence assessment and the final rating round tabulations for each recommendation.

For additional information on the Appropriateness Criteria methodology and other supporting documents, please go to the ACR website at <https://www.acr.org/Clinical-Resources/Clinical-Tools-and-Reference/Appropriateness-Criteria>.

Appropriateness Category Names and Definitions






Appropriateness Category Name	Appropriateness Rating	Appropriateness Category Definition
Usually Appropriate	7, 8, or 9	The imaging procedure or treatment is indicated in

		the specified clinical scenarios at a favorable risk-benefit ratio for patients.
May Be Appropriate	4, 5, or 6	The imaging procedure or treatment may be indicated in the specified clinical scenarios as an alternative to imaging procedures or treatments with a more favorable risk-benefit ratio, or the risk-benefit ratio for patients is equivocal.
May Be Appropriate (Disagreement)	5	The individual ratings are too dispersed from the panel median. The different label provides transparency regarding the panel's recommendation. "May be appropriate" is the rating category and a rating of 5 is assigned.
Usually Not Appropriate	1, 2, or 3	The imaging procedure or treatment is unlikely to be indicated in the specified clinical scenarios, or the risk-benefit ratio for patients is likely to be unfavorable.

Relative Radiation Level Information

Potential adverse health effects associated with radiation exposure are an important factor to consider when selecting the appropriate imaging procedure. Because there is a wide range of radiation exposures associated with different diagnostic procedures, a relative radiation level (RRL) indication has been included for each imaging examination. The RRLs are based on effective dose, which is a radiation dose quantity that is used to estimate population total radiation risk associated with an imaging procedure. Patients in the pediatric age group are at inherently higher risk from exposure, because of both organ sensitivity and longer life expectancy (relevant to the long latency that appears to accompany radiation exposure). For these reasons, the RRL dose estimate ranges for pediatric examinations are lower as compared with those specified for adults (see Table below). Additional information regarding radiation dose assessment for imaging examinations can be found in the ACR Appropriateness Criteria® [Radiation Dose Assessment Introduction](#) document.

Relative Radiation Level Designations

Relative Radiation Level*	Adult Effective Dose Estimate Range	Pediatric Effective Dose Estimate Range
0	0 mSv	0 mSv
	<0.1 mSv	<0.03 mSv
	0.1-1 mSv	0.03-0.3 mSv
	1-10 mSv	0.3-3 mSv
	10-30 mSv	3-10 mSv
	30-100 mSv	10-30 mSv

*RRL assignments for some of the examinations cannot be made, because the actual patient doses in these procedures vary as a function of a number of factors (e.g., region of the body exposed to ionizing radiation, the imaging guidance that is used). The RRLs for these examinations are designated as "Varies."

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Disclaimer

The ACR Committee on Appropriateness Criteria and its expert panels have developed criteria for determining appropriate imaging examinations for diagnosis and treatment of specified medical condition(s). These criteria are intended to guide radiologists, radiation oncologists and referring physicians in making decisions regarding radiologic imaging and treatment. Generally, the complexity and severity of a patient's clinical condition should dictate the selection of appropriate imaging procedures or treatments. Only those examinations generally used for evaluation of the patient's condition are ranked. Other imaging studies necessary to evaluate other co-existent diseases or other medical consequences of this condition are not considered in this document. The availability of equipment or personnel may influence the selection of appropriate imaging procedures or treatments. Imaging techniques classified as investigational by the FDA have not been considered in developing these criteria; however, study of new equipment and applications should be encouraged. The ultimate decision regarding the appropriateness of any specific radiologic examination or treatment must be made by the referring physician and radiologist in light of all the circumstances presented in an individual examination.

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