Prenatal
Aneuploidy *screening*

Washington State Department of HEALTH

Provider Fact Sheet

Aneuploidy Screening and Neural Tube Defects

Prenatal cfDNA(Non-Invasive Prenatal Screening), Ultrasound, First Trimester Screening, Second Trimester Screening, First and Second Trimester Ultrasound, Combined Sequential Screening, QuadScreen, Maternal Serum Screen (MSS), Maternal Serum Alpha Fetal Protein (MS-AFP)

Task Force Recommendations

There are many options for prenatal aneuploidy and neural tube defect (NTD) screening. Patients should be provided with information and resources to make an informed decision and plan for their pregnancy.

All screening and testing options for aneuploidy should be recommended to all pregnant patients, for all pregnancies, regardless of prior pregnancy or family history, and regardless of pregnant person's age at delivery.

Individuals with a prior pregnancy or a family history of genetic conditions should speak with a genetics professional before the provider orders genetic testing or screening.

Providers should review accuracy, false positive rates,and insurance coverage for the various screening methods, and be prepared to answer patient questions regarding different screening options.

Genetic screening and testing are not substitutes for ultrasounds and neural tube screening.

To learn more about prenatal genetic screening and additional resources, visit:
doh.wa.gov/prenatal-genetics-for-providers

Video Resources:

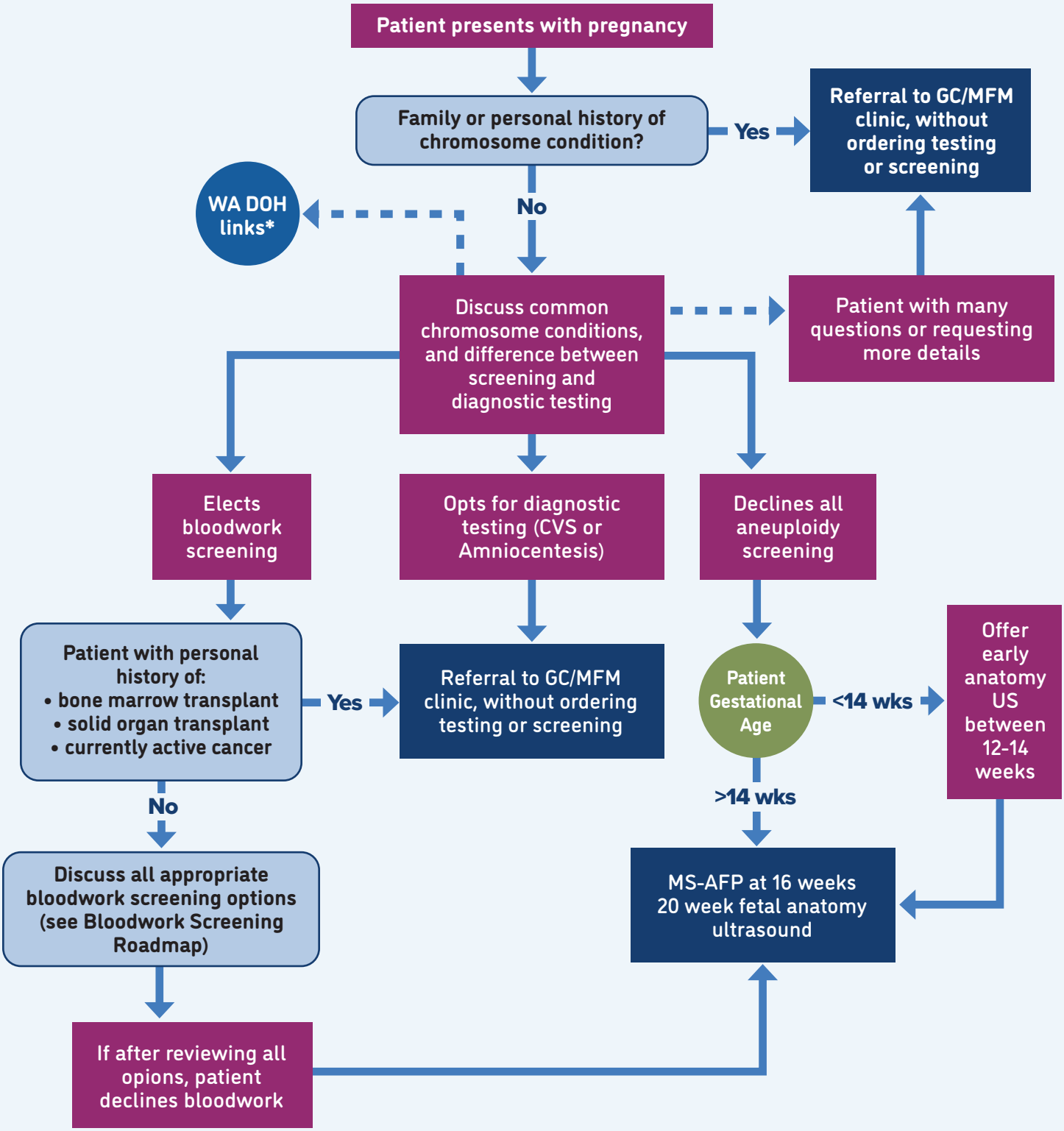
Prenatal Genetic Testing: Making an Informed Decision

(PLAY ALL)

- How to decide about prenatal testing
- What conditions are commonly tested for during pregnancy?
- Prenatal cell-free DNA testing
- Prenatal ultrasound
- Amniocentesis
- Chorionic Villus Sampling (CVS)
- Genetic Carrier Screening

Chapter 246-680 WAC:
Chapter 246-680 of the Washington State Administrative Code (WAC) Stateis the Rule that establishes the standards for screening and diagnostic procedures for prenatal diagnosis of congenital disordersof the fetus under RCW 48.21.244, 48.44.344, and 48.46.375; and to establish criteria and timelines regarding the availability and use of prenatal tests for health care providers to share with pregnant women and couples as required under RCW 70.54.220.

Prenatal Aneuploidy Screening
DETECTION ROADMAP



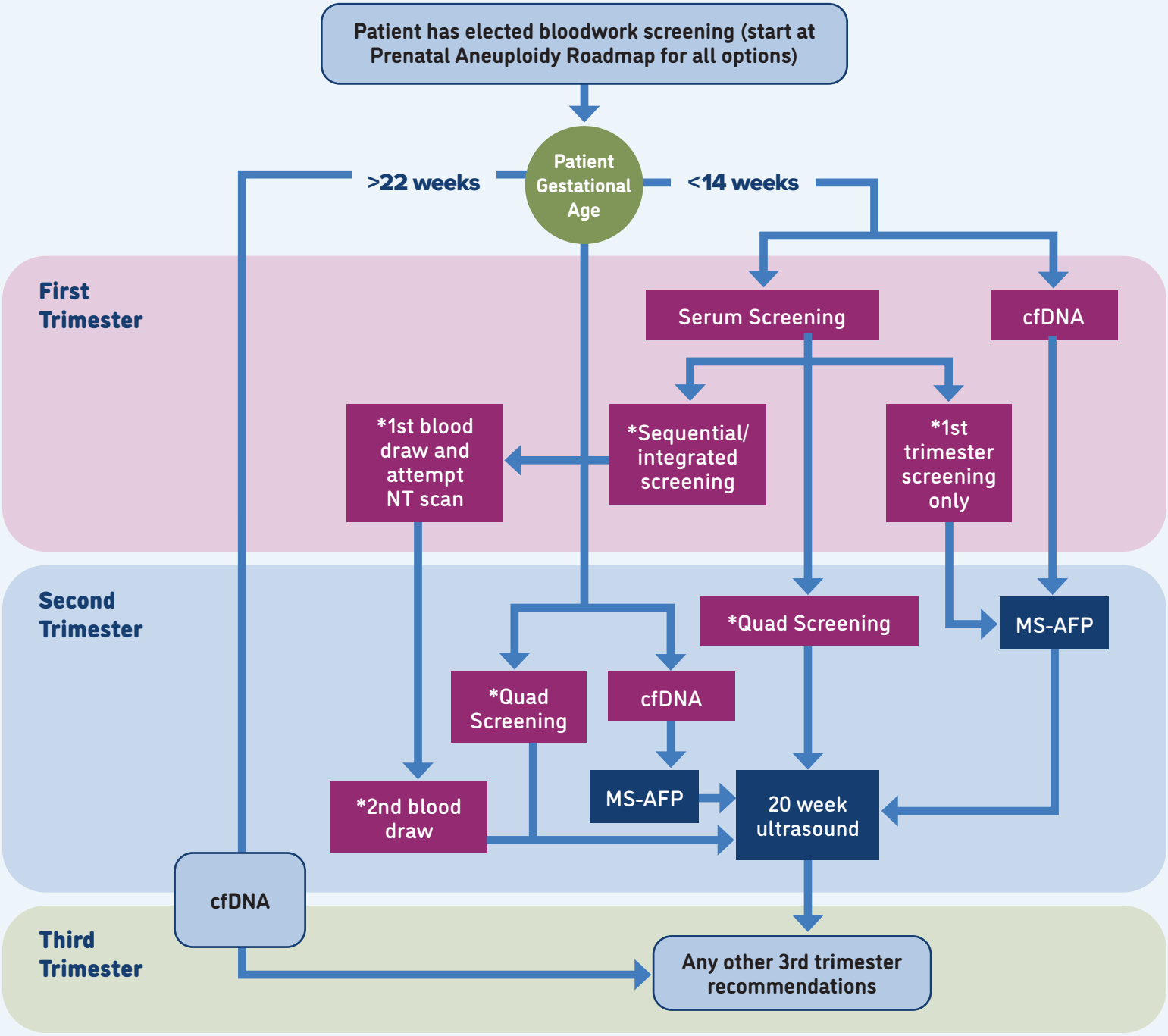
*Prenatal Genetic Screening Information for Patients

"Screening vs. Diagnostic Testing"
(https://doh.wa.gov/public-health-healthcare-providers/healthcare-professions-and-facilities/patient-care-resources/genetic-services/prenatal-genetic-screening-information-patients)

Created in partnership with the Prenatal Genetics Task Force

Prenatal Aneuploidy Screening
BLOODWORK SCREENING ROADMAP

Provider Fact Sheet
Aneuploidy Screening



*do not offer when patient is 35 years or older at delivery
**lab specifics may vary

ADDITIONAL INFORMATION

cfDNA (NIPS)

- Screen for Trisomy 21, Trisomy 18, and Trisomy 13
- Can also include screening for X/ Y chromosomes.
- 93-99% detection rate
- <1% false positive rate
- Insurance may/may not cover, patients w/ Medicaid cannot be balance billed, and self-pay options are typically available.
- Drawn any time after 10 weeks**

Serum Screening

- Screen for Trisomy 21 and Trisomy 18
- 80-90% detection
- 5% false positive rate
- Insurance likely to cover
- 1st trimester blood draws between 11w** and 13w6d**
- 2nd trimester blood draws between 15**-21w6d**

MS-AFP

- Screens for open neural tube defects
- 90% detection rate anencephaly
- 80% detection rate for open neural tube defects
- Insurance is likely to cover
- Drawn between 15w**-21w6d**

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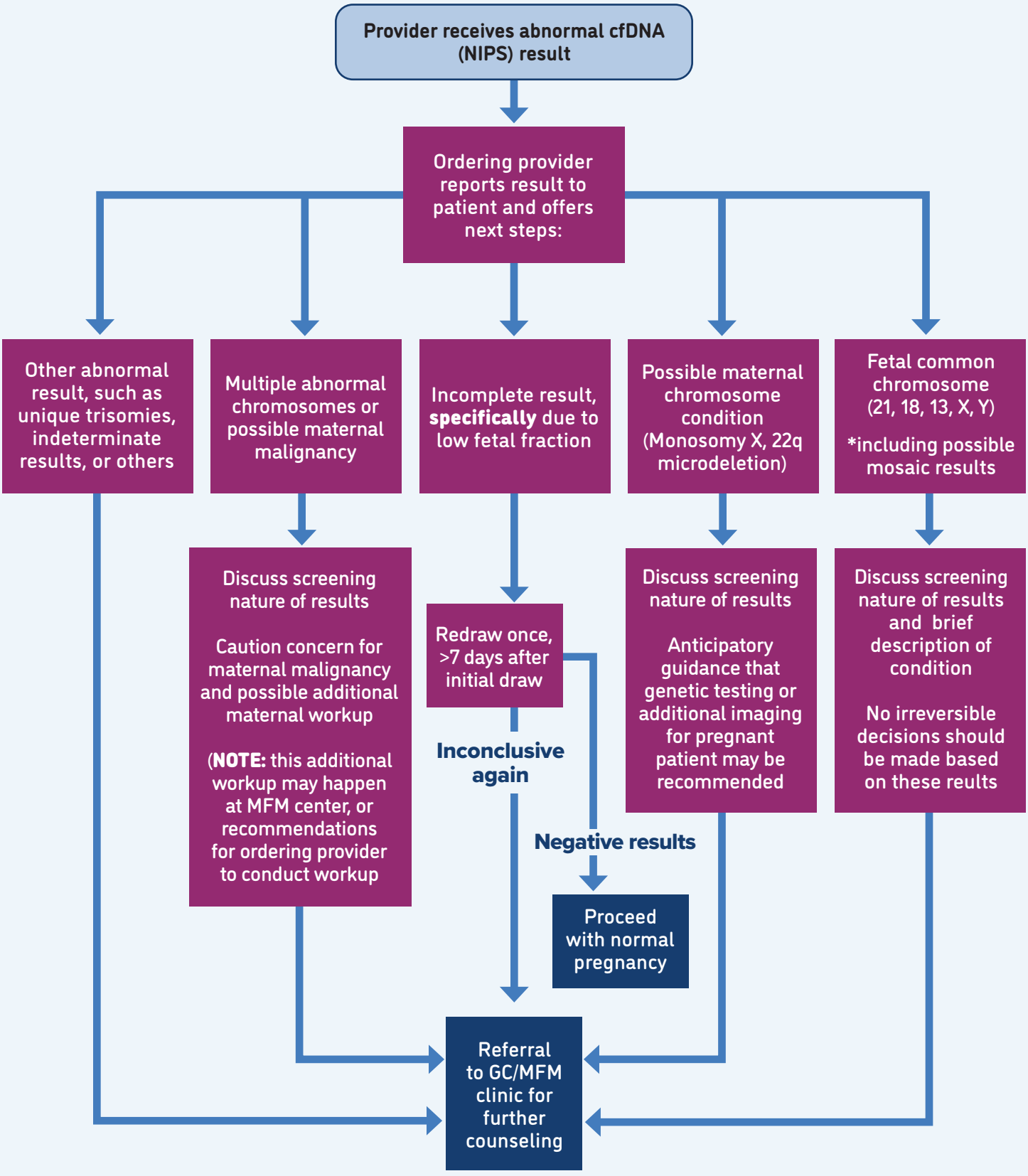

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Current Prenatal Genetic Testing Guidelines

As recommended by the American College of Obstetricians and Gynecologists (ACOG), American College of Medical Genetics and Genomics (ACMG), National Society of Genetic Counselors (NSGC), Society for Maternal-Fetal Medicine (SMFM), American Academy of Family Physicians (AAFP).

Prenatal Type Test	ACOG	ACMG	NSGC	SMFM	AAFP
Combined Guidelines				**Co-published with ACOG-same recommendations as ACOG**	
Maternal Serum Maker	(2016) Recommends: • first trimester combined between 10 and 13 weeks • second trimester screening between 15 and 22 weeks	(2009) Recommends offering to pregnant patients as a screen between 15-20 weeks if they undergo first trimester screening and/or CVS	(2012) Recommends maternal serum to all pregnant patients	(2016) Recommends: • first trimester combined between 10 and 13 weeks • second trimester screening between 15 and 22 weeks	(2014) Recommends the current maternal serum analytes: • alpha-fetoprotein • AFP • hCG • unconjugated estriol Counseling patients about the risks and benefits of screening is important

Current Prenatal Genetic Testing Guidelines (Continued)

Prenatal Type Test	ACOG	ACMG	NSGC	SMFM	AAFP
Combined Guidelines				**Co-published with ACOG-same recommendations as ACOG**	
Prenatal cfDNA	(2023) Recommends that prenatal genetic screening and diagnostic testing options should be discussed and offered to all pregnant patients regardless of maternal age or risk of chromosomal abnormality. Offered anytime from 10 weeks on through the duration of the pregnancy	(2022) Recommends NIPS over traditional screening methods for all pregnant patients with: • singleton gestation for fetal trisomies 21,18, and 13 • twin gestations • singleton gestation to screen for fetal SCA • 22q11.2 deletion syndrome Informing pregnant patients that noninvasive prenatal screening is available. Offered with appropriate pre-and posttest counseling and a referral to trained genetics professionals	(2022) CfDNA screening can be offered to screen for common chromosome abnormalities, like trisomy 21 (Down syndrome), trisomy 18, and trisomy 13 in singleton pregnancies. • Both traditional serum screening and cfDNA are options, but cfDNA has higher sensitivity and specificity. Patients should receive counseling to decide. • CfDNA can be considered for some twin pregnancies as well, with modified positive predictive values. Additional genetic counseling should be provided in this case. • Low risk cfDNA results indicate lowered risk for the targeted trisomies but do not guarantee an unaffected pregnancy. Ultrasound should still be offered.	(2023) Recommends that prenatal genetic screening and diagnostic testing options should be discussed and offered to all pregnant patients regardless of maternal age or risk of chromosomal abnormality. Offered anytime from 10 weeks on through the duration of the pregnancy	(2022) Recommends noninvasive prenatal screening as the most sensitive and specific modality to screen for trisomy 13, 18, and 21

Current Prenatal Genetic Testing Guidelines (Continued)

Prenatal Type Test	ACOG	ACMG	NSGC	SMFM	AAFP
Combined Guidelines				**Co-published with ACOG-same recommendations as ACOG**	
Early Imaging	(2022) Recommends at least one standard exam during your pregnancy, performed at 18–22 weeks of pregnancy. Some may have an ultrasound exam in the first trimester of pregnancy. This is not standard since it is too early to see many of the fetus's detailed limbs and organs. An ultrasound exam done this early is used to do the following: • Estimate gestational age • Help screen for certain genetic disorders • Count the number of fetuses • Check the fetus's heart rate • Check for ectopic pregnancy	(2008) Recommends first trimester screening between 11-14 weeks as an effective and reliable screening test for Down syndrome and trisomy 18 Benefits of first trimester screening: • earlier identification of the pregnancy at risk for fetal aneuploidy and anatomic defects • cardiac anomalies • the option of earlier diagnosis by chorionic villus sampling, if available	None	(2022) Recommends at least one standard exam during your pregnancy, performed at 18–22 weeks of pregnancy. Some may have an ultrasound exam in the first trimester of pregnancy. This is not standard since it is too early to see many of the fetus's detailed limbs and organs. An ultrasound exam done this early is used to do the following: • Estimate gestational age • Help screen for certain genetic disorders • Count the number of fetuses • Check the fetus's heart rate • Check for ectopic pregnancy	(2014) Recommends performing early ultrasonography if the patient has: • irregular cycles or bleeding • if the patient is uncertain of the timing of her last menstrual period • if there is a discrepancy in the size of her uterus compared with the gestational age Ultrasound dating is considered accurate to within four to seven days in the first trimester, 10 to 14 days in the second trimester, and 21 days in the third trimester

Prenatal cfDNA Detections

Test Statistic	Number of Studies	Result (%) (95% CI)
Trisomy 21		
Sensitivity	17	98.80 (97.81-99.34)
Specificity	14	99.96 (99.92-99.98)
Trisomy 18		
Sensitivity	6	98.83 (95.45-99.71)
Specificity	7	99.93 (99.83-99.97)
Trisomy 13		
Sensitivity	7	100 (0-100)
Specificity	8	99.96 (99.92-99.98)

Source: Genetics in Medicine - Performance of NIPS Table
(https://www.gimjournal.org/action/showFullTableHTML?isHtml=true&tableId=tbl1&pii=S1098-3600%2822%2900714-6)

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DOH 344-087 November 2023 CS