Here are some more highlights from the ACMG recommendations:

- Providing up-to-date, balanced, and accurate information early in gestation to optimize patient decision
  making, independent of the screening approach used.
- Laboratories work with public health officials, policymakers, and private payers to make NIPS, including
  the pre- and post-test education and counseling, accessible to all pregnant women.
- Allowing patients to select diagnostic or screening approaches for the detection of fetal aneuploidy and/or genomic changes that are consistent with their personal goals and preferences.
- Informing all pregnant women that diagnostic testing (CVS or amniocentesis) is an option for the detection
  of chromosome abnormalities and clinically significant CNVs.
- Informing all pregnant women that NIPS is the most sensitive screening option for traditionally screened
  aneuploidies (i.e., Patau, Edwards, and Down syndromes).
- Referring patients to a trained genetics professional when an increased risk of aneuploidy is reported after
  NIPS.
- Offering diagnostic testing when a positive screening test result is reported after NIPS.
- Providing accurate, balanced, up-to-date information, at an appropriate literacy level when a fetus is diagnosed with a chromosomal or genomic variation in an effort to educate prospective parents about the condition of concern. These materials should reflect the medical and psychosocial implications of the diagnosis.
- Laboratories should provide readily visible and clearly stated DR, SPEC, PPV, and NPV for conditions being
  screened, in pretest marketing materials, and when reporting laboratory results to assist patients and providers
  in making decisions and interpreting results.
- Laboratories should not offer screening for Patau, Edwards, and Down syndromes if they cannot report DR, SPEC, and PPV for these conditions.
- NIPS to screen for autosomal aneuploidies other than those involving chromosomes 13, 18, and 21 should NOT be offered.
- Offering diagnostic testing for a no-call NIPS result due to low fetal fraction if maternal blood for NIPS was drawn at an appropriate gestational age. A repeat blood draw is NOT appropriate.

- Offering aneuploidy screening other than NIPS in cases of significant obesity.
- All laboratories should include a clearly visible fetal fraction on NIPS reports.
- All laboratories should establish and monitor analytical and clinical validity for fetal fraction.
- All laboratories should specify the reason for a no-call when reporting NIPS results.
- Informing patients that a no-call result may be due to long stretches of homozygosity, which could be due to either UPD or parental consanguinity.
- Referring patients to a trained genetics professional when a no-call result suspicious for UPD or parental consanguinity is received.
- Offering diagnostic testing with CMA when a no-call result is obtained after NIPS due to possible UPD or parental consanguinity.
- Informing all pregnant women, as part of pretest counseling for NIPS, of the availability of the expanded use of screening for sex chromosome aneuploidies.
- Providers should make efforts to deter patients from selecting sex chromosome aneuploidy screening for the sole purpose of biologic sex identification in the absence of a clinical indication for this information.
- Informing patients about the causes and increased possibilities of false-positive results for sex chromosome aneuploidies as part of pretest counseling and screening for these conditions. Patients should also be informed of the potential for results of conditions that, once confirmed, may have a variable prognosis (e.g., Turner syndrome) before consenting to screening for sex chromosomes.
- Referring patients to a trained genetics professional when an increased risk of sex chromosome aneuploidy is reported after NIPS.
- Offering diagnostic testing when a positive screening test result is reported after screening for sex chromosome aneuploidies.
- Providing accurate, balanced, up-to-date information and materials at an appropriate literacy level when a fetus is diagnosed with a sex chromosome aneuploidy in an effort to educate prospective parents about the specific condition. These materials should reflect medical and psychosocial implications for the diagnosis.

- Laboratories include easily recognizable and highly visible DR, SPEC, PPV, and NPV for each sex chromosome
  aneuploidy when reporting results to assist patients and providers in making decisions and interpreting results.
- Laboratories should not offer screening for sex chromosome aneuploidies if they cannot report DR, SPEC, PPV, and NPV for these conditions.
- Informing all pregnant women of the availability of the expanded use of NIPS to screen for clinically relevant
  CNVs when the following conditions can also be met:
    - Obstetric care providers should discuss with their patients the desire for prenatal screening as opposed to diagnostic testing (i.e., CVS or amniocentesis).
    - Obstetric care providers should discuss with their patients the desire for maximum fetal genomic information through prenatal screening.
    - Obstetric care providers should inform their patients of the higher likelihood of false-positive and false-negative results for these conditions as compared to results obtained when NIPS is limited to common aneuploidy screening.
    - Obstetric care providers should inform their patients of the potential for results of conditions that, once confirmed, may have an uncertain prognosis.
- Referring patients to a trained genetics professional when NIPS identifies a CNV.
- Offering diagnostic testing (CVS or amniocentesis) with CMA when NIPS identifies a CNV.
- Providing accurate, balanced, up-to-date information at an appropriate literacy level when a fetus is diagnosed
  with a CNV in an effort to educate prospective parents about the condition of concern. These materials should reflect the medical and psychosocial implications of the diagnosis.
- Laboratory requisitions and pretest counseling information should specify the DR, SPEC, PPV, and NPV of each CNV screened. This material should state whether PPV and NPV are modeled or derived from clinical
  utility studies (natural population or sample with known prevalence).
- Laboratories include easily recognizable and highly visible DR, SPEC, PPV, and NPV for each CNV screened
  when reporting laboratory results to assist patients and providers in making decisions and interpreting results.
  Reports should state whether PPV and NPV are modeled or derived from clinical utility studies (natural
  population or sample with known prevalence). When laboratories cannot report

specific DR, SPEC, PPV, and NPV, screening for those CNVs should not be performed by that laboratory.

- NIPS to screen for genome-wide CNVs is NOT recommended. If this level of information is desired, then diagnostic testing (e.g., chorionic villous sampling or amniocentesis) followed by CMA is recommended.
- In pregnancies with multiple gestations and/or donor oocytes, testing laboratories should be contacted
  regarding the validity of NIPS before it is offered to the patient as a screening option.
- Informing patients of the possibility of identifying maternal genomic imbalances and that this possibility
  depends on the specific methodology used.
- Referring patients to a trained genetics professional when NIPS identifies maternal genomic imbalances.
- Offering aneuploidy screening other than NIPS for patients with a history of bone marrow or organ transplantation from a male donor or donor of uncertain biologic sex.
- Discussing the possibility of discordant fetal biologic sex if maternal blood transfusion was performed <4 weeks prior to the blood draw for NIPS.
- Laboratories provide patient-specific PPV when reporting positive test results.
- Laboratories provide population-derived PPV when reporting positive results in cases in which patient-specific
  PPV cannot be determined due to unavailable clinical information.
- Laboratories provide modeled PPV when reporting positive results for which neither patient-specific nor
  population-derived PPV are possible.
- Providers use validated online calculators to provide patient-specific PPV when results from NIPS are positive
  to facilitate clear and accurate communication with patients.
- Incorporating laboratory-specific DR and SPEC to provide clear and patient-specific information when
  using validated online calculators.