SDx Labs

Prenactive™ Noninvasive Prenatal Screen Plus

Screen for a broader range of chromosomal aneuploidies

The all-chromosome option Prenactive Noninvasive Prenatal Screen Plus will provide information about trisomies for all chromosomes, giving you and your patients a broader range of information than the Prenactive Noninvasive Prenatal Screen.

This option within the Prenactive Noninvasive Prenatal Screen Plus allows screening for rare autosomal trisomies that may occur, especially in the presence of an abnormal ultrasound. Guidelines recommend invasive diagnostic follow-up for those patients with ultrasound anomalies; however, for those who decline invasive diagnostic follow-up, the all-chromosome screening could be an option. Chromosomal aneuploidies, in general, may lead to varying degrees of structural defects, and developmental and intellectual disabilities.[1]

Chromosomal aneuploidies identified with this screen may be representative of the chromosomal make up of every fetal cell (full fetal aneuploidy), some fetal cells (fetal mosaicism), placental cells only (confined placental mosaicism), or some maternal cells (maternal mosaicism).

The clinical significance of rare chromosome aneuploidies is variable and depends on the specific finding and which cells are involved. Patients with a high-risk NIPS result should be offered further detailed counseling and diagnostic testing such as chorionic villus sampling or amniocentesis.[2]

Extensive options for more personalized screening

The Prenactive Noninvasive Prenatal Screen Plus offers the following testing options:

- Aneuploidy of chromosomes 21, 18, and 13 (trisomy 21, 18, and 13) is always included
- Sex chromosome aneuploidies (monosomy X, XXX, XXY, and XYY) are included if requested; fetal sex (XX or XY) will be reported if no sex chromosome aneuploidy is detected
- Aneuploidy (trisomy) of all chromosomes, including sex chromosome aneuploidies
- Select microdeletions such as 1p36 deletion, 4p- (Wolf-Hirschhorn syndrome), 5p- (cri-du-chat syndrome), 15q11.2 (Prader-Willi syndrome/ Angelman syndrome), and 22q11.2 deletion (DiGeorge) are included if requested
- Available for singleton pregnancies >10 weeks gestation

Clear, concise results

Results from the Prenactive Noninvasive Prenatal Screen Plus are reported as “High-Risk: Aneuploidy Detected” or “Low-Risk: No Aneuploidy Detected.” Results for chromosomes 21, 18, 13, X, and Y will continue to be reported individually. Results for the remaining chromosomes are reported collectively. A specific chromosomal aneuploidy will be reported in the event of a “High-Risk: Aneuploidy Detected” result. Results for a positive microdeletion syndrome will be reported as “Results consistent with a microdeletion in a certain genomic region.”

Analytical validation for all chromosomes

	Sensitivity	95% CI	Specificity	95% CI
All autosomes	98.7%	96.1%–99.6%	99.95%	99.62%–>99.99%

The cohort was composed of samples for which high coverage sequencing data identified them as either affected by autosomal aneuploidy (N=189) or otherwise exhibiting normal diploidy (N=1330). The data for these samples were then analyzed at the normal level of sequencing coverage to establish the sensitivity and specificity of the improved algorithm.

Limitations of the screen

Noninvasive prenatal screening (NIPS) based on cell-free DNA analysis from maternal blood is a screen; it is not diagnostic. Test results must not be used as the sole basis for diagnosis. Further genetic counseling and confirmatory diagnostic testing is necessary prior to making any irreversible pregnancy decision. Limited data exist regarding the clinical outcome of pregnancies with rare autosomal trisomies, especially for those detected through NIPS. Health care providers should be aware of the limitations of the screen and understand that limited outcome data may confound pregnancy counseling and management.

About SDxLabs

SDxLabs delivers noninvasive diagnostic and women's health solutions that support proactive healthcare decisions. Our services focus on early detection and actionable results that enable effective treatments and improved patient outcomes. SDxLabs is committed to empowering patients, providers, and the community with access to relevant health services and unmatched service.

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References

1. Gardner RJM, Sutherland GR, Shaffer LG. Chromosome Abnormalities and Genetic Counseling. New York, NY: Oxford University Press; 2012.
2. Practice bulletin no. 163: Screening for fetal aneuploidy. Obstet Gynecol. 2016;127(5):e123-137.