cfDNA started as a screening test for Down syndrome, but over time the list of conditions that can be screened for is expanding. Most labs now provide screening for Down syndrome, trisomy 18, and trisomy 13, as well as the sex chromosomes (X and Y). Down syndrome, trisomy 18, and trisomy 13 are all caused by an extra chromosome.

Screening for sex chromosomes can help predict the sex of the baby, but it can also screen for extra or missing sex chromosomes. These sex chromosome differences can vary widely from mild with no notable physical or developmental differences to severe and life-limiting in rare cases. There are four more common sex chromosome differences that are screened for: Monosomy X/Turner syndrome (45,X), Triple X (47,XXX), Klinefelter syndrome (47,XXY), and Jacobs syndrome (47,XYY).

Some labs have recently started offering screening for a set of genetic conditions called microdeletion syndromes. Microdeletions are when a small piece of a chromosome is missing, rather than an entire chromosome. There are currently no medical guidelines in place to support using cfDNA to screen for microdeletion syndromes, but there may be instances where this information could be helpful. Some microdeletion conditions that labs are screening for are:

- 22q11 deletion syndrome
- 1p36 deletion syndrome
- Angelman syndrome
- Prader-Willi syndrome
- Cri-du-chat
- Wolf-Hirschhorn syndrome
- Jacobsen syndrome
- Langer-Giedion syndrome

Because there is great variation in the conditions that may be screened for, it is important to discuss these options with your provider to make a decision that is best for you and your family.

Click here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

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