

Fetal Genetic Testing

What's my risk?

| Maternal age at delivery | Risk of trisomy 21 | Risk of any chromosomal abnormality |
|--------------------------|--------------------|-------------------------------------|
| 20 | 1/1667 | 1/526 |
| 22 | 1/1429 | 1/500 |
| 24 | 1/1250 | 1/476 |
| 26 | 1/1176 | 1/476 |
| 27 | 1/1111 | 1/455 |
| 28 | 1/1053 | 1/435 |
| 29 | 1/1000 | 1/417 |
| 30 | 1/952 | 1/385 |
| 31 | 1/909 | 1/385 |
| 32 | 1/769 | 1/322 |
| 33 | 1/602 | 1/286 |
| 34 | 1/485 | 1/238 |
| 35 | 1/378 | 1/192 |
| 36 | 1/289 | 1/156 |
| 37 | 1/224 | 1/127 |
| 38 | 1/173 | 1/102 |
| 39 | 1/136 | 1/83 |
| 40 | 1/106 | 1/66 |
| 41 | 1/82 | 1/53 |
| 42 | 1/63 | 1/42 |
| 43 | 1/49 | 1/33 |
| 44 | 1/38 | 1/36 |
| 45 | 1/30 | 1/21 |
| 46 | 1/23 | 1/16 |
| 47 | 1/18 | 1/13 |
| 48 | 1/14 | 1/10 |
| 49 | 1/11 | 1/8 |

Data modified from maternal age-specific rates by Hook EB (1981) Rates of chromosome abnormalities at different maternal ages. *Obstetrics Gynecology* 58: 282; and Hook EB, Cross PK, and Schreinemacher DM (1983) Chromosomal abnormality rates at amniocentesis and in live-born infants. *Journal of the American Medical Association* 249: 2034.

Cell Free Fetal DNA "MaterniT21"

- 9-10 weeks +
 - Fetal cells circulating in maternal blood
 - Screens for Down Syndrome, Trisomies 18, 13
 - Detection rates 99% (False Positive 0.1%)



- 11-14 weeks
 - US (Nuchal Translucency) and blood work (hCG and PAPP-A)
 - Screen for Down Syndrome, Trisomy 18
 - Detection rates 83% (False Positive 5%)

First Trimester Screen

- 15-22 weeks
 - Blood work (AFP, estriol, hCG, inhibin A)
 - Screens for Down Syndrome, Trisomy 18, Spina Bifida
 - Detection rates 70-81% (False Positive 5%)

Quad Screen



AFP Screen

- 16 weeks
 - 1 in 1,000 pregnancies effected
 - Spontaneous failure of neural tube closure during early embryonic development
 - Open body wall defects detected
 - Measures Alphafetoprotein (AFP)

Cystic Fibrosis

Spinal Muscular Atrophy

Parental Carrier Screening

- Rare pulmonary disease
- 1:2,500 births
- Most common genetic disorder
- CF causes mucus build up in the lungs and pancreas.
- Results in infertility
- Rare neuromuscular disease
- 1:6,000 births
- SMA can cause profound breathing issues, severe muscular weakness, and irreversible loss of nerves in the spine.
- Onset of weakness can start at birth up to adulthood.
- Major cause of death in infancy.

