

# Prenatal Cell-free DNA Screening

## What does it test for?

### Common trisomies

#### Trisomy 21 (Down syndrome)

- Characteristic facial features
- Mild to moderate cognitive delay
- May have heart and/or other medical conditions

#### Trisomy 18 (Edwards syndrome)

- Multiple birth defects
- Severe cognitive delay
- High mortality rate in infancy

#### Trisomy 13 (Patau syndrome)

- Multiple birth defects
- Severe cognitive delay
- High mortality rate in infancy

### Microdeletions

- Small missing pieces of chromosomes
- Signs and symptoms vary
- Most results in growth and/or development delays

### Fetal sex

cfDNA testing can detect the presence of the X and the Y chromosome and can predict the sex of the fetus.

X

Y

### Rh Factor

- Can determine if fetus is Rh positive or negative
- Useful for Rh sensitive women
- May need to be ordered as a separate test

### Sex chromosome testing

#### XXY (Klinefelter syndrome)

#### XO (Turner syndrome)

#### Others (depends on laboratory)

- Mild or no effect on development
- Possible puberty and fertility issues
- Possible learning difficulties

### Other conditions and the future

- Less common trisomies
- Single-gene disorders
- Complex disorders
- Whole genome/exome sequencing

To learn more:

Prenatal cell-free DNA screening: <https://www.pathlms.com/ashg/course/4595>

National Down Syndrome Society: [www.ndss.org](http://www.ndss.org)

International Trisomy 13/18 Alliance: [www.trisomy.org](http://www.trisomy.org)

Unique: [www.rarechromo.org](http://www.rarechromo.org)

Chromosome Disorder Outreach: [www.chromodisorder.org](http://www.chromodisorder.org)