

## A new screening option for Down syndrome, trisomy 18 and open neural tube defects

genzyme

### If I am “screen positive” what additional testing is available?

Your doctor may offer you one of the following diagnostic procedures:

**Chorionic villi sampling (CVS)** is a procedure that takes a small amount of the tissue from the developing placenta. The tissue is then sent to a laboratory for chromosome analysis. CVS can be done between 10 and 12 weeks of pregnancy. CVS is associated with a small risk of miscarriage.

**Amniocentesis** is a procedure that takes a small amount of the fluid that surrounds the baby. The fluid is then sent to a laboratory to test for chromosome abnormalities and open neural tube defects. An amniocentesis is usually done around the 16th week of pregnancy. Amniocentesis is also associated with a small risk of miscarriage, however the risk is lower than that for CVS.

### About Genzyme Genetics

Genzyme Genetics has been a world leader in genetic testing and counseling services for 25 years.

This brochure is provided by Genzyme Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web site at [www.genzyme.com](http://www.genzyme.com).

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### SequentialScreen

- SequentialScreen provides early answers for women with the highest chances of having a baby with Down syndrome and trisomy 18.
- SequentialScreen is a two-part test that provides most women with two answers:
  - An early, preliminary result
  - A final, more complete result

### SequentialScreen detects approximately:

#### Part 1

- 70% of the babies with Down syndrome
- 80% of the babies with trisomy 18

#### Part 2

- 90% of babies with Down syndrome
- 90% of babies with trisomy 18
- 80% of babies with open neural tube defects



# SequentialScreen

## What is a screening test?

A screening test is a safe way to learn more about your baby's chances of having certain birth defects. A screen is meant to identify people at high risk, not to provide a diagnosis. This brochure discusses SequentialScreen, a new screening option for Down syndrome, trisomy 18 and open neural tube defects.

## What is SequentialScreen?

SequentialScreen is a two-part screen and the only screen that can provide you with two answers – an early, preliminary result in the first trimester and a final result in the second trimester.

## What is Down syndrome?

Down syndrome is caused by an extra chromosome #21. This extra chromosome causes mental and physical abnormalities. About 1 in 800 babies is born with Down syndrome. The risk of Down syndrome gradually increases with the age of the mother, but can occur at any age.

## What is trisomy 18?

Trisomy 18 is caused by an extra chromosome #18. This extra chromosome causes severe mental and physical problems. About 1 in 6,500 babies is born with trisomy 18. Only 1 out of 10 babies with trisomy 18 lives past the age of one year. As with Down syndrome, the risk of trisomy 18 gradually increases with the age of the mother.

## What are open neural tube defects?

The neural tube, which forms very early in pregnancy, eventually develops into the baby's brain and spinal cord. If this tube does not close completely, an opening remains along part of the baby's spine or head. This can lead to paralysis and other physical and/or mental problems. Open neural tube defects occur in 1 out of 1,000 live births. The risk of having a child with an open neural tube defect does not increase with the age of the mother.

## How does SequentialScreen work?

### Part 1

Between your 10<sup>th</sup> and 14<sup>th</sup> week of pregnancy, a small amount of blood and a special ultrasound measurement will be taken. SequentialScreen will use information from your blood and ultrasound measurement to provide you with your risk of having a baby with Down syndrome or trisomy 18. If you're at an increased risk, your doctor will offer diagnostic testing to confirm the results of your screen.

### Part 2

If you are not identified as someone at increased risk, you will come back between your 15<sup>th</sup> and 22<sup>nd</sup> week for a second blood test. The second part of this screen will use the information collected at both times in your pregnancy and provide you with a final screening result. The screen will tell you what your risk is of having a baby with Down syndrome, trisomy 18 or an open neural tube defect.

## What are the possible test results, and what do they mean?

Shortly after you have your blood drawn, your doctor will receive your screen result.

### Part 1

#### Final result pending second trimester sample

This result means that your pregnancy is not in the highest risk group for Down syndrome or trisomy 18. You will be asked to complete the second part of this screen.

#### Screen positive

This result means that there is an increased risk of your baby having Down syndrome or trisomy 18. The next step is to talk with your doctor and/or genetic counselor about your results and diagnostic testing options.

### Part 2

#### Screen negative

This result means that there is a low risk of your baby having Down syndrome, trisomy 18 or an open neural tube defect. However, in approximately 10% of pregnancies, these birth defects will not be detected through screening.

#### Screen positive

This result means that there is an increased risk of your baby having Down syndrome, trisomy 18 or an open neural tube defect. The next step is to talk with your doctor and/or genetic counselor about your results and diagnostic testing options.

