PRENATAL SCREENING
IS IT RIGHT FOR YOU?

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ABOUT THIS PROGRAM

This booklet and the accompanying video are intended for women who are considering prenatal screening for Down syndrome, trisomy 18 and neural tube defects.

Did you receive the video?
You can watch it online at **http://bit.ly/dh_pdp**

This program is *not* intended for women who

- Have already had prenatal screening for Down syndrome, trisomy 18 and neural tube defects in this pregnancy.
- Have already had chorionic villus sampling or amniocentesis in this pregnancy.
- Are past 22 weeks of pregnancy.

ABOUT THIS DECISION

Pregnancy is an exciting time filled with many important decisions. One decision is whether or not to have prenatal screening for Down syndrome, trisomy 18 and neural tube defects. While all pregnant women are offered this screening, it should not be considered a routine test. The results may lead expectant parents to face very difficult decisions about their pregnancy.

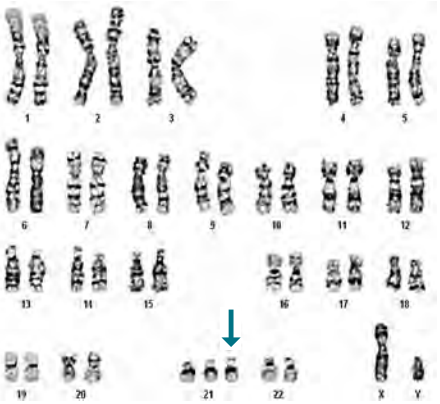
In this booklet, you will find information about Down syndrome, trisomy 18 and neural tube defects and the screening and testing options available to you at Dartmouth-Hitchcock.

"I chose to have prenatal screening because I was scared, and I wanted confirmation that everything was okay." —Sarra

"For me personally, the screening test can put a level of anxiety there that may or may not need to be there and may lead to testing that doesn't need to be done." —Kristy

"I needed to know in advance, so I needed to have the prenatal screening for my own peace of mind." —Cassandra

WHAT IS DOWN SYNDROME?

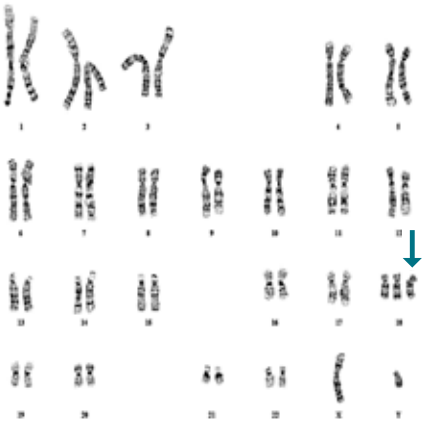


Down syndrome, also called trisomy 21, results from an extra chromosome. Individuals with Down syndrome have varying degrees of intellectual disability. They may also have birth defects, such as heart and intestinal conditions. Life expectancy is currently over 50 years of age.

Down syndrome occurs in about 1 in 700 live births. The chance of having a child with Down syndrome increases with the age of the mother (see table, *page 10*).

"The things they do are going to happen later. They're going to walk at a later age; they're going to talk at a later age. In most respects, these developmental events are going to occur, and it's kind of like a book in which the pages are turning a little more slowly." —Carl Cooley, MD

WHAT IS TRISOMY 18?



Like Down syndrome, trisomy 18 also results from an extra chromosome. Individuals with trisomy 18 have severe intellectual and physical disabilities. They may have birth defects of almost any organ system, including the heart, brain, and kidneys. Most babies with trisomy 18 are stillborn or do not survive beyond infancy.

Trisomy 18 occurs in about 1 in 4000 live births. As with Down syndrome, the chance of having a child with trisomy 18 increases with the age of the mother (see table, *page 11*).

"Children with trisomy 18 usually have enough of a motor disability that they're not able to walk unassisted. [They] are not able to speak, but they do understand and comprehend much more than they can say. Overall, when studies have been performed of developmental skills of older children—five to ten years of age—again, usually the skills are somewhere before the twelve-month level." —John Carey, MD

Live Birth Prevalence of Down Syndrome According to Maternal Age

Maternal Age at Delivery	Down syndrome	
	1 in	*per 10,000*
16	1509	7
17	1504	7
18	1497	7
19	1488	7
20	1476	7
21	1461	7
22	1441	7
23	1415	7
24	1381	7
25	1339	7
26	1285	7
27	1219	8
28	1139	8
29	1045	8
30	937	9
31	819	10
32	695	11
33	571	12
34	455	14
35	352	18
36	266	22
37	199	28
38	148	37
39	111	50
40	85	67
41	67	89
42	54	116
43	45	148
44	39	182
45	35	217
46	31	250
47	29	281
48	27	309
49	26	332

Figures are rounded to the nearest whole number

Live Birth Prevalence of Trisomy 18 According to Maternal Age

Maternal Age at Delivery	Trisomy 18	
	1 in	*per 10,000*
16	9090	1
17	9090	1
18	9090	1
19	9090	1
20	9090	1
21	9090	1
22	9090	1
23	9090	1
24	9090	1
25	8333	1
26	8333	1
27	8333	1
28	8333	1
29	7692	1
30	7143	1
31	6667	2
32	5882	2
33	5263	2
34	4348	2
35	5371	3
36	2703	4
37	2041	5
38	1449	7
39	1031	10
40	735	14
41	532	19
42	398	25
43	311	32
44	253	40
45	214	47
46	188	53
47	170	59
48	157	34
49	149	67

Figures are rounded to the nearest whole number

WHAT ARE NEURAL TUBE DEFECTS?

ILLUSTRATION : A.D.A.M.

Neural tube defects are birth defects of the brain and spinal cord that occur early in pregnancy. The two most common neural tube defects are spina bifida and anencephaly. Individuals with spina bifida may have problems with walking and bowel and bladder control issues. Babies with anencephaly die shortly after birth.

Neural tube defects occur in about 1 in 1500 live births. Taking folic acid prior to pregnancy and during the first trimester can help to prevent many neural tube defects.

"Children with this condition can do very well. It's important to realize that families who raise a child with spina bifida are going to face medical and developmental challenges…They will be involved in an ongoing process where specialists monitor their child's care, and varying surgical procedures will be offered over time to help ameliorate or prevent difficulties." —Ardis Olson, MD

WHAT IS PRENATAL SCREENING?

Screening tests involve measuring certain pregnancy-related proteins in your blood; many screening tests also use information from an ultrasound. Prenatal screening only tells you if your baby may have Down syndrome, trisomy 18 or a neural tube defect. Further tests would be needed to tell you for sure.

Prenatal screening results

Prenatal screening results are reported as either screen positive or screen negative.

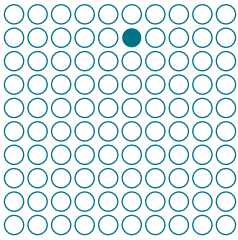
- A screen positive result means a high risk for the baby to have one of the conditions.
- A screen negative result means a low risk.

The lab will use a cut-off to decide if the result is screen positive or screen negative. Thus, a screen positive result means that the risk for your baby to have the condition is at or above the cut-off. A screen negative result means the risk is below the cut-off.

Most labs will report a specific risk for each condition. For example, a 1 in 10 risk means that for every 10 women who have this result, one will have a child with the condition, while the other nine will not.

A 1 in 10 risk.

Similarly, a 1 in 100 risk means that for every 100 women who have this result, one will have a child with the condition, while the other 99 will not.



A 1 in 100 risk.

These are the 4 possible outcomes that you can have with a screening test:

- *False positive:* The result shows a high risk for Down syndrome, trisomy 18 or a neural tube defect when the baby does not have the condition.

- *True positive:* The result shows a high risk for Down syndrome, trisomy 18 or a neural tube defect and the baby does have the condition.

- *False negative:* The result shows a low risk for Down syndrome, trisomy 18 or a neural tube defect when the baby does have the condition.

- *True negative:* The result shows a low risk for Down syndrome, trisomy 18 or a neural tube defect and the baby does not have the condition.

  The majority of screening test results are true negatives.

"In our case, we had a very high number that really made us nervous and stressed out more than probably what we needed to be." —Nick

"Be aware that there are false positives and the test…isn't fool proof. It doesn't tell you everything, and sometimes… even babies who are…born with disabilities don't come up in the screening." —Clarissa

WHAT HAPPENS IF MY SCREENING TEST IS POSITIVE?

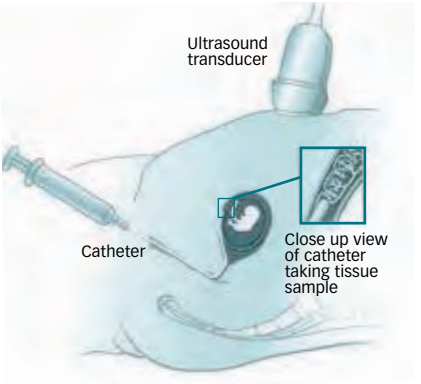
If your screening test is positive, you will be offered further screening and diagnostic testing. The screening tests include a targeted fetal anatomy ultrasound and non-invasive prenatal testing. The diagnostic tests include chorionic villus sampling and amniocentesis; these can tell you for sure but have a risk of miscarriage.

Targeted Fetal Anatomy Ultrasound:
A targeted fetal anatomy ultrasound is done between 18 and 20 weeks to evaluate the baby's growth and development. This ultrasound is reviewed by a maternal-fetal medicine specialist. There is no risk of miscarriage with this test. While a normal result makes it less likely that the baby has Down syndrome or trisomy 18, it cannot tell you for sure if the baby has either of these conditions. Most neural tube defects will be diagnosed by ultrasound. This test is available at Dartmouth-Hitchcock Medical Center in Lebanon, Dartmouth-Hitchcock Manchester and Dartmouth-Hitchcock Nashua.

Non-Invasive Prenatal Testing (NIPT):
During pregnancy, there are chromosome fragments from both the mother and the baby in the mother's blood; this is known as cell-free DNA. NIPT is a blood test that measures the amount of cell-free DNA from chromosome 21 and chromosome 18. If there is an increased amount, then there is a high risk that the baby has Down syndrome or trisomy 18. NIPT can be done starting at 10 weeks. It is only available to women whose pregnancies are known to be at an increased risk for a chromosome disorder; among these women, NIPT detects about 99% of babies with Down syndrome and about 97% of babies with trisomy 18. Chorionic villus sampling or amniocentesis would be needed to confirm a high risk result. This test is available at Dartmouth-Hitchcock Medical Center in Lebanon, Dartmouth-Hitchcock Manchester and Dartmouth-Hitchcock Nashua.

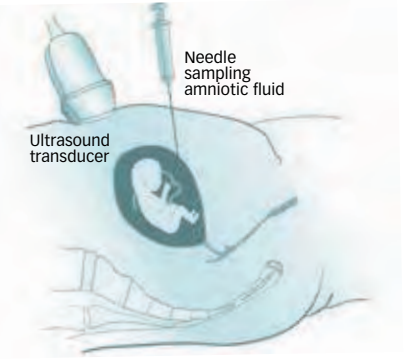
Chorionic Villus Sampling (CVS):

CVS involves obtaining a small sample from the placenta either by inserting a needle through the abdomen or by inserting a catheter through the vagina and cervix. CVS is done between 11 and 13 weeks and can diagnose Down syndrome and trisomy 18. It cannot test for neural tube defects. The risk of miscarriage is up to 10 in 1,000 (1%). This test is only available at Dartmouth-Hitchcock Manchester.



Amniocentesis:

Amniocentesis involves obtaining a small amount of amniotic fluid by inserting a needle through the abdomen and into the uterus. Amniocentesis is done beginning at 15 weeks gestation and can diagnose Down syndrome, trisomy 18 and neural tube defects. The risk of miscarriage is up to 3 in 1,000 (0.3%). This test is available at Dartmouth-Hitchcock Medical Center in Lebanon, Dartmouth-Hitchcock Manchester and Dartmouth-Hitchcock Nashua.



IS SCREENING RIGHT FOR ME?

Although prenatal screening may seem like a routine test, the decision to have the test is not something you should take lightly. Most prenatal screening results are reassuring, but it is important to think about how you might feel if you have a positive result.

When deciding if screening is right for you, ask yourself the following questions. It may be helpful to share your answers with your prenatal care provider.

- Am I prepared to deal with the worry that I may feel if I have a positive screening result?
  ❒ Yes ❒ No ❒ I'm not sure

- If I have a positive screening result, would I consider having a CVS or an amniocentesis to get a definite answer? (Remember: CVS and amniocentesis both have a risk of miscarriage.)
  ❒ Yes ❒ No ❒ I'm not sure

- Would I consider terminating my pregnancy if I find out that my baby definitely has Down syndrome, trisomy 18 or a neural tube defect?
  ❒ Yes ❒ No ❒ I'm not sure

- If I decide to continue my pregnancy, would knowing that my baby has one of these conditions help me to prepare for his or her birth?
  ❒ Yes ❒ No ❒ I'm not sure

- If I decide to continue my pregnancy, would I want to have time during my pregnancy to consider an adoption plan?

  ❒ Yes ❒ No ❒ I'm not sure

- Would I rather find out after my baby's birth if he or she has one of these conditions?

  ❒ Yes ❒ No ❒ I'm not sure

"I'd tell folks to tell themselves, 'What...am I going to do differently with this information? Am I prepared to do the next step?' So, if the next step is something you don't want to do, then don't start down the path." —Mike

WHAT ARE MY SCREENING TEST OPTIONS?

If you decide to have screening, there are several options. The following information is about the screening tests that are available at Dartmouth-Hitchcock. The names and details of each test may be different at other centers.

If you are less than 14 weeks pregnant, you will need to choose one of the following tests:

- First Trimester Screen
- Integrated Screen
- Sequential Screen
- Serum Integrated Screen
- Quad Marker Screen

If you are already more than 14 weeks pregnant, the Quad Marker Screen is your only option.

Here are some medical terms that you will need to understand as you read about these tests:

Nuchal Translucency: The fluid-filled space under the skin of the baby's neck as measured by ultrasound.

Screen Positive Rate: The number of test results that are above the screen cut-off. This rate includes both false positives and true positives.

Detection Rate: The number of affected pregnancies that will have a test result above the screen cut-off.

FIRST TRIMESTER SCREEN

When is it done?

First Trimester Screening is done between 11 and 13 weeks.

How is it done?

First Trimester Screening combines a nuchal translucency ultrasound with a blood test. The blood test measures three pregnancy-related proteins: PAPP-A, hCG, and inhibin (DIA).

	Down syndrome	Trisomy 18	Neural tube defects
Cut-off	Risk of 1 in 220	Risk of 1 in 100	--
Detection rate	84 out of 100	82 out of 100	--
Screen positive rate	5 out of 100	< 1 out of 100	--

< means "less than"

What are the advantages?

- This test gives the earliest risk estimate for Down syndrome and trisomy 18.

What are the disadvantages?

- This test has the highest screen positive rate for Down syndrome.
- This test has lower detection rates than most of the other screening tests.
- This test does not give a risk estimate for neural tube defects; however, this may be done by a separate blood test at a later time.

INTEGRATED SCREEN

When is it done?

- Integrated Screening is a two-step process. Step one is done between 11 and 13 weeks. Step two is done between 15 and 20 weeks.

How is it done?

- Step one combines a nuchal translucency ultrasound with a blood test. This blood test measures a pregnancy-related protein called PAPP-A. *There are no results reported from step one.*
- Step two is second blood test. This blood test measures four pregnancy-related proteins: AFP, estriol (uE3), hCG and inhibin (DIA). Results are reported after this step.

	Down syndrome	Trisomy 18	Neural tube defects
Cut-off	Risk of 1 in 250	Risk of 1 in 100	AFP of 2.0 MoM
Detection rate	90 out of 100	89 out of 100	85 out of 100
Screen positive rate	3 out of 100	< 1 out of 100	2 out of 100

MoM = multiples of the median; < means "less than"

What are the advantages?

- This test has the highest detection rate for Down syndrome and trisomy 18.
- Two-step screening tests, like Integrated Screening, have a lower screen positive rate than one-step screening tests.

What are the disadvantages?

- There are no results until after 15 weeks (after the second blood draw).

SEQUENTIAL SCREEN

When is it done?

- For most women, Sequential Screening is a two-step process. Step one is done between 11 and 13 weeks. Step two is done between 15 and 20 weeks.

How is it done?

- Step one combines a nuchal translucency ultrasound with a blood test. This blood test measures a pregnancy-related protein called PAPP-A. *There are no results reported from step one unless the risk for Down syndrome is at least 1 in 40. Women who receive a result after step one will not go on to step two.*
- Women who do not receive a result from step one will go on to step two. Step two is a second blood test. This blood test measures four pregnancy-related proteins: AFP, estriol (uE3), hCG, and inhibin (DIA).

		Down syndrome	Trisomy 18	Neural tube defects
Cut-off	Step one	Risk of 1 in 40	Risk of 1 in 100	--
	Step two	Risk of 1 in 200	Risk of 1 in 100	AFP of 2.0 MoM
Detection rate		89 out of 100	88 out of 100	85 out of 100
Screen positive rate		3 out of 100	< 1 out of 100	2 out of 100

MoM = multiples of the median; < means "less than"

What are the advantages?

- This test provides an early result for pregnancies with the highest risk for Down syndrome.
- Two-step screening tests, like Sequential Screening, have a lower screen positive rate than one-step screening tests.

What are the disadvantages?

- Most women will not get results until after 15 weeks (after the second blood draw).

SERUM INTEGRATED SCREEN

When is it done?

- Serum Integrated Screening is a two-step process. Step one is done between 11 and 13 weeks. Step two is done between 15 and 20 weeks.

How is it done?

- Step one is a blood test. This blood test measures a pregnancy-related protein called PAPP-A. *There are no results reported from step one.*
- Step two is a second blood test. This blood test measures four pregnancy-related proteins: AFP, estriol (uE3), hCG and inhibin (DIA). Results are reported after this step.

	Down syndrome	Trisomy 18	Neural tube defects
Cut-off	Risk of 1 in 150	Risk of 1 in 100	AFP of 2.0 MoM
Detection rate	85 out of 100*	89 out of 100	85 out of 100
Screen positive rate	3 out of 100	< 1 out of 100	2 out of 100

MoM = multiples of the median; < means "less than"

*Detection rate only applies to women whose dating has been confirmed by ultrasound. If an ultrasound has not been done, the detection rate for Down syndrome is 80 out of 100.

What are the advantages?

- This is the best test for women who do not have access to a nuchal translucency ultrasound.
- Two-step screening tests, like Serum Integrated Screening, have a lower screen positive rate than one-step screening tests.

What are the disadvantages?

- This test has a lower detection rate for Down syndrome than other two-step screening tests.
- There are no results until after 15 weeks (after the second blood draw).

QUAD MARKER SCREEN

When is it done?

- Quad Marker Screening is done between 15 and 20 weeks.

How is it done?

- Quad Marker Screening involves only one blood test. The blood test measures four pregnancy-related proteins: AFP, estriol (uE3), hCG and inhibin (DIA).

	Down syndrome	Trisomy 18	Neural tube defects
Cut-off	Risk of 1 in 150	Risk of 1 in 100	AFP of 2.0 MoM
Detection rate	76 out of 100	68 out of 100	85 out of 100
Screen positive rate	4 out of 100	< 1 out of 100	2 out of 100

MoM = multiples of the median; < means "less than"

What are the advantages?

- This test is available to women who are greater than 14 weeks and did not have earlier screening.

What are the disadvantages?

- This test has the lowest detection rate for Down syndrome and trisomy 18.

Comparing Screening Test Options

	First Trimester Screen	Integrated Screen	Sequential Screen	Serum Integrated Screen	Quad Marker Screen
Detection rates:					
Down syndrome	84 out of 100	90 out of 100	89 out of 100	85 out of 100	76 out of 100
Trisomy 18	82 out of 100	89 out of 100	88 out of 100	89 out of 100	68 out of 100
Neural tube defect	--	85 out of 100	85 out of 100	85 out of 100	85 out of 100
Benefits:					
Highest detection rate		✔			
Lowest screen positive rate		✔	✔	✔	
Early results	✔		✔		
Follow-up testing options:					
Targeted Fetal Morphology Ultrasound	✔	✔	✔	✔	✔
Non-Invasive Prenatal Testing	✔	✔	✔	✔	✔
CVS	✔		✔		
Amniocentesis	✔	✔	✔	✔	✔

PRENATAL DIAGNOSIS PROGRAM CONTACT INFORMATION

Dartmouth-Hitchcock Medical Center
One Medical Center Drive
Lebanon, NH 03756
(603) 650-6025

Dartmouth-Hitchcock Manchester
Dartmouth-Hitchcock Specialty Care at Bedford Medical Park
5 Washington Place
Bedford, NH 03110
(603) 695-2902

Dartmouth-Hitchcock Nashua
2300 Southwood Drive
Nashua, NH 03063
(603) 577-4324

FOR MORE INFORMATION

Prenatal Testing and Genetic Counseling

- Dartmouth-Hitchcock
  http://patients.dartmouth-hitchcock.org/obstetrics/prenatal_testing_genetic_counseling.html
- National Society of Genetic Counselors
  http://www.nsgc.org

Decision aides

- Ottawa Personal Decision Guide
  http://decisionaid.ohri.ca/decguide.html
- Should I have an amniocentesis?
  http://patients.dartmouth-hitchcock.org/health_information/health_encyclopedia/aa103080
- Should I have CVS?
  http://patients.dartmouth-hitchcock.org/health_information/health_encyclopedia/tb1905

Down syndrome resources

- The National Down Syndrome Society
  http://www.ndss.org
- Northern New England Down Syndrome Congress
  http://www.nnedsc.org

Trisomy 18 resources

- Support Organization for Trisomy (SOFT)
  http://www.trisomy.org
- Trisomy 18 Organization
  http://www.trisomy18.org

Neural tube defect resources

- Spina Bifida Association
  http://www.spinabifidaassociation.org

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