

# Prenatal Diagnosis

Public  
Education  
Pamphlet



The Society of  
Obstetricians and  
Gynaecologists of  
Canada

## PRENATAL DIAGNOSIS

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About 2 to 3% of babies born have some type of major birth defect. The risk of some problems, due to abnormal separation of genetic material (chromosomes), increases with the mother's age. About 50% of the time these types of problems are due to Down Syndrome, which is a third copy of chromosome 21 (Trisomy 21). The other half of chromosomal anomalies are caused by a variety of problems. Many of these chromosomal problems result in a severely affected baby or one which does not survive even to delivery. The incidence of Down Syndrome and other chromosomal problems increases with age. At 35 years the risk is about 1:200. It is at this age that direct testing of fetal cells is offered.



### Amniocentesis

An amniocentesis is a test in which the cells that are floating in the fluid surrounding the fetus are examined for the chromosomal pattern of the fetus. An amniocentesis is offered at about 15 weeks gestation and results are available two to three weeks later. Amniocentesis carries a risk of losing the pregnancy of about 1:200 or 0.5%.

### Chorionic Villus Sampling

Some centers also offer Chorionic Villus Sampling (CVS) which can provide almost the same results earlier in the pregnancy. The pregnancy loss rate is slightly higher with CVS than amniocentesis.

Many women who are eligible for amniocentesis or Chorionic Villus Sampling prefer not to take the added risk of a miscarriage. These women, and younger women who wish some assurance, can be offered maternal serum screening (MSS). This test is possible because some substances made by the fetus cross over into the mother's blood stream. This is usually done at 15 to 16 weeks by taking a small amount of blood from a vein in the mother's arm.

Amniocentesis is offered to women who appear to be at increased risk after maternal serum screening.

### Single Screen Test

A Single Screen looks for alpha fetoprotein (AFP). If this were high, it would suggest an increased risk of spina bifida (spina bifida can also be picked up on an 18 week screening ultrasound, about 95% of the time). A low level of AFP suggests an

increased risk of Down Syndrome. This test will pick up approximately 30% of babies with Down Syndrome.

### Double Screen Test

More information can be provided by a Double Screen which measures both AFP and Human Chorionic Gonadotrophin (HCG). The combination of these two tests will pick up approximately 60% of infants with Down Syndrome.

### Triple Screen Test

A Triple Screen measuring AFP, HCG, and Estriol, a hormone produced by the placenta, improves the pick up rate to approximately 70%.

A low or high result does not necessarily mean that the fetus has an abnormality. It can mean that the fetus is older or younger than thought to be, that there are two or more fetuses, or there could be other conditions relating to abnormal levels.

In addition to Down Syndrome and neural tube defects, such as spina bifida, a small number of other chromosomal problems can also be picked up by this sort of testing. Availability of these tests varies from province to province. Testing can be done but is not always covered by Provincial Departments of Health. In that case, patients would have to assume the cost of testing. All provinces currently cover AFP. The Double Screen is covered in some provinces, and if not, would cost approximately \$40. The Triple Screen is covered in some provinces, and if not, would cost approximately \$80.

Chorionic Villus Sampling is offered around 10 weeks. It involves putting a needle into the womb and taking a sample of tissue which will develop into the placenta (afterbirth).

An amniocentesis would usually be offered around 15 weeks. This test involves inserting a needle into the sac of fluid that surrounds the fetus.

Chorionic Villus Sampling and amniocentesis can be offered to:

- pregnant women who will be 35 or older on their due date;
- couples who have had a child with a birth defect or have a family history of certain birth defects;
- pregnant women with other abnormal test results (such as ultrasound or MSS).

In the case of Chorionic Villus Sampling, it would take one to two weeks for the results to be reported. The couple with an affected fetus would be faced with making a decision regarding continuing the pregnancy at approximately 12 weeks gestation.

In the case of amniocentesis, it takes approximately two to three weeks for the results to be reported. A couple with an affected fetus diagnosed with amniocentesis would then be faced with

making a decision regarding continuing the pregnancy at approximately 18 to 19 weeks.

Both Chorionic Villus Sampling and amniocentesis are covered by Provincial Health Plans. However, availability varies from region to region.

### Maternal Serum Screen

Maternal Serum Screen (single screen, double screen or triple screen) is usually done at 15 to 16 weeks. Reports are available

in one to two weeks. This testing cannot tell if the baby has or does not have a chromosomal problem but instead gives an estimate of risk, such as the risk of a 20 year-old or the risk of a 40 year-old. The couple is then faced with deciding whether or not to have more definitive testing and again must wait several weeks for the report of this testing. When an abnormal result is reported, you should discuss your options with your care giver. This timing also applies to a problem identified on the screening ultrasound that is done at about 18 weeks. The latest time in a pregnancy when a termination could be performed varies from province to province. It is generally between 20 and 24 weeks at most.

A decision regarding prenatal testing should be made early in a pregnancy. Most provinces offer genetic counselling to aid in the decision for invasive prenatal testing. A couple should make a decision regarding Maternal Serum Screen after considering what they would do if the results suggested that there was a problem.

RISK OF CHROMOSOMAL ABNORMALITIES IN LIVE-BORN INFANTS		
Maternal Age (yr)	Risk for Down Syndrome	Total Risk for Chromosomal Abnormalities*
20	1/1,667	1/526
21	1/1,667	1/526
22	1/1,429	1/500
23	1/1,429	1/500
24	1/1,250	1/476
25	1/1,250	1/476
26	1/1,176	1/476
27	1/1,111	1/455
28	1/1,053	1/435
29	1/1,000	1/417
30	1/952	1/384
31	1/909	1/384
32	1/769	1/323
33	1/625	1/286
34	1/500	1/238
35	1/385	1/192
36	1/294	1/156
37	1/227	1/127
38	1/175	1/102
39	1/137	1/83
40	1/106	1/66
41	1/82	1/53
42	1/64	1/42
43	1/50	1/33
44	1/38	1/26
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

\*47, xxx excluded for ages 20-32 (data not available). Modified from the following sources: Hook EB, Cross PK, Schreinemachers DM. Chromosomal abnormality rates at amniocentesis and in live-born infants. JAMA 1983;249(15):2034-38 (ages 33-49). This table was reprinted with the permission of JAMA. Hook EB. Rates of chromosomal abnormalities at different maternal ages. Obstet Gynecol 1981;58(3):282-85

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The Society of Obstetricians and Gynaecologists of Canada  
 780 Echo Drive, Ottawa, Ontario K1S 5R7  
 Tel: 613-730-4192  
 Fax: 613-730-4314  
 1-800-561-2416  
[www.sogc.org](http://www.sogc.org)

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