



# Genetics and Birth Defect: Questionnaire

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1.	Will you be 35 years or older when your baby is due?	YES	NO
2.	<b>IF</b> you or your spouse are <i>African American</i> , Mediterranean, or Asian: have you been screened for sickle cell trait or thalassemia and <b>found to be positive</b> ?	YES	NO
3.	Do you work with raw meats or eat raw meat or fish?	YES	NO
4.	Have you ever had chicken pox?	YES	NO
5.	Do you work with any chemicals or potentially toxic agents?	YES	NO
	If yes, please list:		
6.	Do you own cats?	YES	NO
	If yes, are they indoor, outdoor or both?		
7.	<b>IF</b> you or your spouse are Jewish, please explain your background:		
8.	Do you use or have you used any of the following during this pregnancy:		
	a. Tobacco	YES	NO
	b. Alcohol	YES	NO
	c. Illegal drugs	YES	NO
	d. Prescription medications	YES	NO
	e. Herbal supplements	YES	NO
9.	Do you have a history of infertility, abortion, stillbirth, or any other serious pregnancy complication?	YES	NO
	If yes, please explain:		

## Have you or any members of your family ever had:

Name of the Disease	YES	NO	Please Explain below
Mental retardation			
Mental illness			
Epilepsy			
Other diseases of the nervous system			
Down syndrome			
Tay-Sachs			
Canavan's Disease			
Huntington's Chorea			
Muscular dystrophy			
Cystic fibrosis			
Hydrocephalus			
Spina bifida			
Clotting disorder			
Hemophilia or bleeding disorder			
Any other chromosomal disorder			
Any other birth defect not listed above?			

*If you have any questions after reading the information on the next page, please discuss with your physician.*

I am interested in having further genetic counseling. \_\_\_\_\_ YES NO

I have completed this questionnaire and have read the comments. \_\_\_\_\_ YES NO

\_\_\_\_\_  
Patient Signature

\_\_\_\_\_  
Date



# Genetics and Birth Defect: About

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While most children are born healthy, approximately 3-5% of children are born with some sort of birth defect. These defects can be very minor or severe and typically develop in the first three months of pregnancy. About 70% of the time, the cause of the defect is not known. However, known causes of birth defects can also include genetics or exposure of the mother to harmful agents or medications.

*Second trimester anomaly screening* involves drawing blood work between weeks 15 and 20 of pregnancy. Different testing can be done, but in our office, we use what is called a “quad screen.” This means that we check the blood for four different hormones. When these levels are combined with other information about the mother, a level of risk can be assessed to the pregnancy of the fetus having one of the following abnormalities:

1. Trisomy 18
2. Neural tube defects (detected in 80% of cases)
3. Down syndrome (detects Down syndrome in 80% of cases)

**NEURAL TUBE DEFECTS** refer to an incomplete closure of the fetal spine that can result in spina bifida or *anencephaly*. 95% of these problems occur in patients without a family history of similar problems. The risk of this defect in the Southeastern United States is 3 infants per 10,000 live births. The risk of this can be decreased with folic acid.

**DOWN SYNDROME** (trisomy 21) risk increases with age. The rates of trisomy 21 over age 30 are as follows (these numbers have been rounded and can vary somewhat):

Mom's Age	Down syndrome	All trisomies	Mom's Age	Down syndrome	All trisomies	Mom's Age	Down syndrome	All trisomies
21	1 in 1,429	1 in 526	31	1 in 909	1 in 384	41	1 in 82	1 in 53
22	1 in 1,429	1 in 500	32	1 in 769	1 in 323	42	1 in 64	1 in 42
23	1 in 1,429	1 in 500	33	1 in 625	1 in 286	43	1 in 50	1 in 33
24	1 in 1,250	1 in 476	34	1 in 500	1 in 238	44	1 in 38	1 in 26
25	1 in 1,250	1 in 476	35	1 in 385	1 in 192	45	1 in 30	1 in 21
26	1 in 1,176	1 in 476	36	1 in 294	1 in 156	46	1 in 23	1 in 16
27	1 in 1,111	1 in 455	37	1 in 227	1 in 127	47	1 in 18	1 in 13
28	1 in 1,053	1 in 435	38	1 in 175	1 in 102	48	1 in 14	1 in 10
29	1 in 1,000	1 in 417	39	1 in 137	1 in 83	49	1 in 11	1 in 8
30	1 in 952	1 in 384	40	1 in 106	1 in 66			

The testing addressed above is only a *screening* test. If the results of this are abnormal, further testing and counseling with possibly a referral to a maternal fetal medicine specialist may be recommended by your physician. Women with an increased risk of having a baby with a birth defect include women who have a family or personal history of birth defects, women who have had a child with a birth defect or genetic problem, and women who will be over the age of 35 at delivery. If you fall into one of these categories or if you are interested in earlier screening, please call or discuss with your provider so that we can refer you to a specialist.