



# Cleveland & Gilchrist

## Genetics and Birth Defect Questionnaire

1. Will you be 35 years or older when your baby is due? YES      NO
2. **IF** you or your spouse are *African American, Mediterranean, or Asian*: have you been screened for sickle cell trait or thalassemia and **found to be positive**? 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. **IF** 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	FREQUENCY _____
b. Alcohol	YES	NO	FREQUENCY _____
c. Illegal drugs	YES	NO	FREQUENCY _____
d. Prescription medications	YES	NO	FREQUENCY _____
e. Herbal supplements	YES	NO	FREQUENCY _____

 If yes, please list what you are using: \_\_\_\_\_
9. Do you have a history of infertility, abortion, stillbirth, or any other serious pregnancy complication? If yes, please explain: YES      NO

Have you or any members of your family ever had:	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

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.