



Cristina Matera, MD, FACOG | Maureen O'Brien Moomjy, MD, FACOG | Jessica Rosenberg Brown, MD, FACOG

## PRECONCEPTION GENETIC QUESTIONNAIRE AND CONSENT FORM

Name \_\_\_\_\_ Date of Birth \_\_\_\_\_

Partner Name \_\_\_\_\_ Date of Birth \_\_\_\_\_

1. Do you, your partner, or anyone in your families have any of these disorders?

Cystic Fibrosis	Yes	No
Muscular Dystrophy	Yes	No
Hemophilia	Yes	No
Huntington's disease	Yes	No
Polycystic Kidney Disease	Yes	No
Neural tube defect (open spine)	Yes	No
Neurofibromatosis	Yes	No
Marfan syndrome	Yes	No

If yes, please indicate the relationship of the affected person to you or your partner: \_\_\_\_\_

2. Do you or your partner have a birth defect or familial disorder not listed above? Yes No  
If yes, please specify:

3. Do you or your partner have a close relative with mental retardation, autism, any birth defect, Fragile X, familial disorder, or a chromosome disorder such as Down Syndrome? Yes No

If yes, please specify the condition and indicate the relationship:

4. In any previous marriage(s) have you or your partner had a child born with a birth defect or had a pregnancy or child diagnosed with Down Syndrome? Yes No

If yes, please specify the defect: \_\_\_\_\_



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5. Have you or your partner in this or any previous marriage had a stillborn child, a second or third trimester pregnancy loss or two or more first trimester miscarriages? Yes No

If yes, please specify:

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6. Did you or your partner have carrier testing for Cystic Fibrosis? Yes No

If yes, please indicate results and state who was tested:

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7. Are you or your partner of Eastern European (Ashkenazi) Jewish, French-Canadian or Cajun ancestry? Yes No  
Have you been screened for Tay-Sach's disease? Yes No  
If Eastern European Ashkenazi Jew, have you been screened for Canavan, Gaucher, Bloom, Fanconi Anemia, Neimann-Pick Disease, Familial Dysautonomia, Mucopolisaccharidosis Type IV, Maple Syrup Urine Disease, Glycogen Storage Disease Type 1A, Nemaline Myopathy, Usher Syndrome Type 1, Lipoamide dehydrogenase deficiency, or Usher Syndrome Type III? Yes No

If yes, please indicate the results and who was tested:

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8. Are you or your partner of Afro-American, Hispanic, or Caribbean ancestry? Yes No  
Have you been screened for sickle cell trait? Yes No

If yes, please indicate the results and who was tested:

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9. Are you or your partner of Mediterranean (Italian, Greek, North African), Asian (Chinese, Indian, or Pakistani), South East Asian (Taiwanese, Vietnamese), or Middle Eastern (Iranian, Turkish, or Egyptian) background? Yes No  
Have you ever been screened for Beta-thalassemia? Yes No

If yes, please indicate results and who was tested:

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10. Are you or your partner of Asian (Chinese, Indian, Pakistani), or Southeast Asian (Vietnamese, Indonesian, North African, Philippine, Malaysian) ancestry? Yes    No  
 Have you been screened for a-thalassemia? Yes    No

If yes, please indicate test results and who was tested:

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I have completed the "Preconception Genetic Questionnaire" and answered the questions to the best of my knowledge.

Based on my answers the following tests were recommended:

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\_\_\_\_\_ A. I/We **agree/decline** (*please circle*) the recommended test(s) at this time.

\_\_\_\_\_ B. Reason(s) for declining the recommended test(s) are as follows: \_\_\_\_\_

\_\_\_\_\_ C. I/We have received a copy of the "Public Information: Genetic Screening and Planning a Family" and understand that it serves as a general and brief description of what can be expected from genetic screening. For a more thorough, detailed, and/or specific explanation, an appointment for genetic counseling is highly recommended.

\_\_\_\_\_ D. A consideration for genetic counseling has been offered to us and we **accept/decline** (*please circle*) at this time.

We/I understand:

- 1) The test(s) is/are for an abnormality in the genes for the disorder(s), using DNA analysis.
- 2) The purpose of testing is to determine a carrier status (unaffected but able to pass the abnormal gene onto a child by inheritance)
- 3) The test(s) is/are for genetic susceptibility ("genetic predisposition") and that the risk of having the disorder(s) may be altered by family history and/or other factors. If the test(s) is/are positive for the disorder(s) or for an increased risk of the disorder(s), you may wish to have further independent testing or have genetic counseling.
- 4) The results of the above test(s) become a part of your medical record and may be made available to individuals/organizations with legal access to your medical record, on a strict "need-to-know" basis, including, but not limited to physicians and nursing staff directly involved in your care, your current



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and future insurance carriers, and others specifically authorized by you to gain access to the medical record.

5) Your medical insurance may not pay for the test, in which case you will be responsible for the bill.

Patient/Wife's Name \_\_\_\_\_ Signature \_\_\_\_\_  
Partner/Husband's Name \_\_\_\_\_ Signature \_\_\_\_\_  
Physician \_\_\_\_\_ Date \_\_\_\_\_