NYU Langone Health

Preconception Genetic Questionnaire

Patient Name: _____ Date of Birth: _____

Partner Name: _____ Date of Birth: _____

1. Do you, your partner, your children, or anyone in your families have a genetic or chromosomal disorder? If yes, please indicate the relationship of the affected person to you or your partner. _____

   Examples of genetic disorders may include (but are not limited to):

   - *Muscular dystrophy (e.g. Duchenne, myotonic dystrophy)*
   - *Bleeding disorder (e.g. hemophilia)*
   - *Neurofibromatosis*
   - *Dwarfism/skeletal dysplasia*
   - *Marfan syndrome*
   - *Polycystic kidney disease*
   - *Huntington's disease*
   - *Cystic fibrosis*
   - *Spinal muscular atrophy*
   - *Intellectual/developmental disability or autism (e.g. Fragile X syndrome, Down syndrome)*
   - *Birth defect (e.g. spina bifida, cleft palate, heart defect)*
   - *Blindness or deafness*
   - *Hereditary cancer syndrome or cancer diagnosed < age 50*
   - *Balanced translocation*

2. In this or any previous relationship, have you or your partner had a pregnancy diagnosed with a chromosome disorder (e.g. Down syndrome) or a birth defect? If yes, please specify the diagnosis. ☐ No ☐ Yes _____

3. In this or any previous relationship, have you or your partner had a stillbirth or more than two (2) miscarriages? If yes, please provide further information. ☐ No ☐ Yes

   _____

4. Please indicate your ancestry/ethnicity (list all countries of origin):

   Self: _____

   Partner: _____

5. Do you or your partner have any Eastern European (Ashkenazi) Jewish ancestry?
   ☐ Self ☐ Partner

6. Do you or your partner have any French-Canadian or Cajun ancestry?
   ☐ Self ☐ Partner

7. Do you or your partner have any African (including African-American), Caribbean, Hispanic, Asian, Middle Eastern, Mediterranean, or Sephardic/Mizrahi Jewish ancestry?
   ☐ Self ☐ Partner

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8. Did you or your partner have carrier testing for any of the following diseases? If yes, please indicate the results and include a copy of your report if possible.

Cystic Fibrosis (CF)	☐ Self	☐ Partner	_____
Spinal Muscular Atrophy (SMA)	☐ Self	☐ Partner	_____
Fragile X	☐ Self	☐ Partner	_____
Sickle Cell Disease	☐ Self	☐ Partner	_____
Beta Thalassemia	☐ Self	☐ Partner	_____
Alpha Thalassemia	☐ Self	☐ Partner	_____
Bloom Syndrome	☐ Self	☐ Partner	_____
Canavan Disease	☐ Self	☐ Partner	_____
Dihydrolipoamide Dehydrogenase Deficiency	☐ Self	☐ Partner	_____
Familial Dysautonomia	☐ Self	☐ Partner	_____
Familial Hyperinsulinism	☐ Self	☐ Partner	_____
Fanconi Anemia Type C	☐ Self	☐ Partner	_____
Gaucher Disease	☐ Self	☐ Partner	_____
Glycogen Storage Disease Type 1A	☐ Self	☐ Partner	_____
Joubert Syndrome Type 2	☐ Self	☐ Partner	_____
Maple Syrup Urine Disease	☐ Self	☐ Partner	_____
Mucolipidosis Type IV	☐ Self	☐ Partner	_____
Nemaline Myopathy	☐ Self	☐ Partner	_____
Niemann-Pick Disease Type A	☐ Self	☐ Partner	_____
Tay-Sachs Disease	☐ Self	☐ Partner	_____
Usher Syndrome Type IF	☐ Self	☐ Partner	_____
Usher Syndrome Type III	☐ Self	☐ Partner	_____
Walker-Warburg Syndrome	☐ Self	☐ Partner	_____

I and my partner have answered the questions to the best of our knowledge. Based on our responses, my physician, Dr. _____ has recommended genetic counseling and the following testing:

_____ ☐ Accept ☐ Decline

_____ ☐ Accept ☐ Decline

_____ ☐ Accept ☐ Decline

My physician listed above has also requested a genetic consult and the following testing be performed before an In Vitro Fertilization (IVF) cycle can be initiated:

_____ ☐ Accept ☐ Decline

_____ ☐ Accept ☐ Decline

_____ ☐ Accept ☐ Decline