WWC OB-GYN
Wellesley Women's Care

Wellesley Women's Care, P.C.

GENETIC TESTING CONSENT

Wellesley Women's Care routinely tests patient(s) for Cystic Fibrosis, Tay sachs, Spinal Muscular Atrophy and if appropriate, Ashkenazi Jewish Panel (for patients of Ashkenazi Jewish descent). A positive test result is an indication that the patient may be an unaffected or affected carrier and may wish to consider further independent testing, consult their physician and/or pursue genetic counseling.

All testing for recessive genetic diseases is optional and may not be covered by your insurance carrier, thus making you responsible for the full cost of testing.

Cystic Fibrosis

Cystic fibrosis (CF) is an autosomal recessive disease that causes thick mucus and other fluids to build up and clog different parts of the body, including the lungs, pancreas, liver, and intestine. It is the most common monogenic disorder in non-Hispanic whites of Northern European descent, with a carrier frequency of 1/24 to 1/25 and birth prevalence of 1 in 2500. Cystic fibrosis is caused by an abnormal gene. To get the disease, people need to get the abnormal gene from both their mother and father. If people get the abnormal gene from only 1 parent, they will not have cystic fibrosis. But they will have a chance of passing on the abnormal gene to their children.

A negative screening test means only that the individual does not carry any of the CF mutations in the screening battery; a negative result thus reduces the likelihood, **but does not eliminate** the possibility, that the individual is a CF carrier.

Spinal Muscular Atrophy

SMA is the most common monogenic cause of infant mortality. SMA results in progressive muscle weakness and atrophy. The incidence of spinal muscular atrophy (SMA) ranges from 4 to 10 per 100,000 live births, and the carrier frequency of disease-causing SMN1 mutations ranges from 1/90 to 1/47.

Ashkenazi Jewish Genetic screening tests

The American College of Obstetricians and Gynecologists (ACOG) Committee on Genetics recommends routinely offering all Ashkenazi Jews carrier screening for four of the most common disorders that are either lethal or associated with significant morbidity: Tay-Sachs disease, Canavan disease, cystic fibrosis, and familial dysautonomia. There is also an expanded panel that includes Bloom Syndrome, familial hyperinsulinism, Fanconi anemia group C, Gaucher disease, glycogen storage disease type I, Joubert syndrome, Maple syrup urine disease, mucolipidosis IV, Niemann Pick disease type A, and Usher syndrome.

- My signature and initials below indicate that I have read the above information and I understand it.
- I have also read or had explained to me the specific diseases tested for, and the specific test(s) I am having, including the test descriptions, principles, and limitations.
- I have had the opportunity to discuss the purposes and possible risks of this testing.
- I know that genetic counseling is available to me before and after the testing.
- I have all the information I want/need and all my questions have been answered.

____ I decline all routine genetic testing including Cystic Fibrosis, Spinal Muscular Atrophy, Tay sachs and Ashkenazi Jewish Panel (for patients of Ashkenazi Jewish Descent).

____ I consent to routine genetic testing including Cystic Fibrosis, Spinal Muscular Atrophy only and wish to decline Tay sachs and Ashkenazi Jewish Panel (for patients of Ashkenazi Jewish Descent).

____ I consent to routine testing for Cystic Fibrosis, Spinal Muscular Atrophy, Tay sachs and wish to decline the Ashkenazi Jewish Panel (for patients of Ashkenazi Jewish descent).

____ I consent to routine testing for Cystic Fibrosis, Spinal Muscular Atrophy, Tay sachs and Ashkenazi Jewish Panel (for patients of Ashkenazi Jewish descent).

____ I consent to Spinal Muscular Atrophy testing.

Patient Signature Date

Patient Name (please print) Date of Birth