

## Needs Assessment Survey Glossary

The following are definitions for terms that we use in our needs assessment survey about breast health and screening for autistic people.

- **Breast biopsy:** A breast biopsy is when doctors take a small sample of breast tissue to make sure that the tissue is normal. Doctors may do a biopsy using a needle through the skin, or by surgery using anesthesia (medications that make you sleep while the biopsy is being done).
- **Breast cancer:** Breast cancer is an illness where the cells in your breast (very small parts of your breast) grow too much inside your body. Breast cancer may start small but can become very serious if it is not found and treated.
- **Breast MRI:** An MRI is a type of body scan that uses giant magnets to take pictures of the inside of your body and look for signs of damage or illness. Breast MRIs use a special type of MRI machine that is made to take pictures of your breasts to look for cancer or other changes. Although breast MRIs don't hurt, the machine can be very loud.
- **Breast ultrasound:** An ultrasound is a type of body scan that uses sound waves to take pictures of the inside of your body and looks for signs of damage or illness. Breast ultrasounds are used to check for cancer or other changes inside the breast.
- **Gender fluid:** A gender fluid person is a person who does not describe themselves as exclusively male or female.
- **Genes:** Genes are tiny parts of your body that tell your body how to grow and work properly. For example, genes control what color your eyes are.
- **Genetic counseling:** Genetic counseling is a kind of healthcare. During genetic counseling appointments an expert called a genetic counselor explains more about genetic testing, helps order genetic testing for you, explains your test results, and helps you make healthcare decisions based on these results.
- **Genetic counselor:** A type of healthcare professional who has training in genetics. Genetic counselors help people understand their risk of cancer and make decisions about genetic testing. After the test results are returned, they help people understand their results and make decisions about how to protect themselves from cancer.
- **Gene mutations (also called pathogenic variants):** These are changes in genes that can increase the risk for certain diseases or conditions such as cancer. Gene mutations are labeled by the name of the gene with the mutation – usually a combination of 3-6 letters or numbers. Some of the more common gene mutations that can cause cancer to run in families include:
  - ATM
  - BRCA1
  - BRCA2
  - BRIP1
  - CDH1
  - CHEK2
  - Lynch syndrome genes (EPCAM, MLH1, MSH2, MSH6 or PMS2)
  - PALB2
  - PTEN
  - RAD51C
  - RAD51D
  - TP53

- Genetic testing: A type of medical test using your blood or saliva. Genetic tests can look at your genes to see if you have any changes that might make you more likely to become sick. Most genetic tests only look at a small portion of the genes in your body.
- Genetic test results: There are 3 main types of results for genetic testing.
  - Positive for a mutation (also called positive for a pathogenic variant). This means that a change was found in one of your genes. A positive test result will include the name of the gene that has the change. Gene names are usually made up of several letters and sometimes numbers. (For example: BRCA1, MSH1, PALB2).
  - Negative for a mutation (also called negative for a pathogenic variant). This means that the test didn't find any changes in the genes that they looked at.
  - Variant of Uncertain Significance (also called a VUS). These are changes in genes that doctors cannot tell if they are harmful or not.
- Lynch syndrome: A type of condition caused by a mutation in one of the five Lynch syndrome genes (EPCAM, MLH1, MSH2, MSH6 or PMS2). Lynch syndrome causes an increased risk for colorectal and other types of cancer and can cause cancer to run in families.
- Mammogram: A mammogram is a type of xray that takes pictures of the inside of your breast and looks for signs of damage or illness.
- Nonbinary: A nonbinary person is a person who does not describe themselves as exclusively male or female.
- Pathogenic variants: Another name for a gene mutation that can increase the risk for cancer or other diseases or conditions.
- Transgender: A transgender person is a person whose current gender is different than the gender listed on their birth certificate.