Genetic Testing for Babies with BPD

BPD is a complex disease with varying causes and outcomes. Prematurity, the causes and complications of premature birth, and treatments after birth all play a role in the development, and severity of BPD. Genetic differences between children probably play some role as well, but researchers have failed to find a single gene or set of genes that is/are the cause of BPD. With that in mind, there are some limited circumstances in which your baby's medical team may recommend genetic testing.

What are genes?

Our bodies are made up of millions of cells. While different types of cells make up different parts of the body (for example, lung cells, skin cells, blood cells, etc.), they all contain almost identical instructions to function correctly. **These instructions are called genes, and a baby inherits different genes from each parent.** Variations in genes are generally referred to as genetic variants -- Some genetic variants are inherited from one or both parents, while others occur new in a given individual. While most genetic variants do not cause human diseases, a small number do; and parents cannot control which genetic variants are passed on to their child.

Structure of genes in the body

Why pursue genetic testing? What can I expect?

If a member of your child's medical team is suspicious that one or more genetic variants may be contributing to your child's medical condition, a clinical genetic test may be recommended. Potential benefits of genetic testing in these circumstances include:

- Identification of specific therapies which may be beneficial
- Information on other, non-genetic, tests which may be helpful in the future
- Having the peace of mind from knowing that your child's medical condition did have a specific identifiable cause -- and knowing the chances of this happening again, if you plan to have another child
- Knowing that certain rare genetic disorders are NOT the cause of your child's medical condition.

In general, there are three main outcomes from genetic testing:

1. **Positive:** The test has detected one of genetic variants which are causing some or all of your child's health problems. These genetic variants are often called "Pathogenic".
2. **Negative:** The test did not detect genetic variants and there cannot explain your child's health problems.
3. **Uncertain:** The test detected genetic variants which may or may not explain some of your child's health problems. Sometimes this uncertainty can be resolved by testing other family members, sometimes the uncertainty can only be resolved by future research. Often, you child's medical team can use knowledge they have about your child to better interpret an uncertain finding for you.

- In most cases, genetic testing is done using a sample of your baby's blood
- In some circumstances, blood or saliva from one or both parents may also be requested as part of testing

Depending on the complexity of the genetic test recommended or the specific results, you may be asked to meet with a medical geneticist or genetic counselor to discuss the risks, benefits and alternatives to the proposed genetic testing.