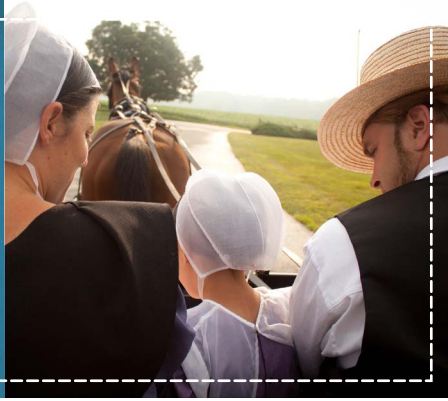
Plain Insight Panel™

Expanded Carrier Testing



Insight into your carrier status

The Plain Insight Panel™ (PIP) is **a genetic test** that can determine carrier status for a wide variety of inherited conditions. The PIP is specific to conditions found in the **Plain communities across the United States** and **currently includes over 1,400 genetic changes (variants) that can affect health.** Some of these genetic variants are associated with conditions commonly managed at the Clinic, while others are very rare and their effect is not immediately obvious. The vast majority of variants on the PIP determine your carrier status for recessive disorders, while a few variants may have personal implications for your health.

Why does carrier status matter?

Most people do not know if they are carriers of any genetic conditions. This is because carriers do not experience any symptoms. Often, they do not have a family history of a disorder. **All people**, Plain and non-Plain, **are carriers for a handful of conditions**. Since many Plain couples have ancestors in common, members of the Plain Community are more likely to be carriers for the same genetic conditions. Only genetic testing will determine this for certain. For this reason, the PIP is best done by couples together.

Carrier status is most significant if both parents are carriers for variants in the **same gene**. In that case, each baby has a 25% chance (1 in 4) of being **affected** with the condition. If we know that a couple is at risk for having a child with a genetic condition, we can diagnose the baby at birth using cord blood. If a targeted treatment exists for a condition, like GA-1 or MSUD, the baby can then be treated before they show symptoms. In many cases, affected babies can start treatment that makes a significant difference in their health, and parents feel prepared for what lies ahead.

More than **1,400** *variants in the Plain Insight Panel™*

ALL *Plain couples should consider testing together*

5 *variants on average are carried by people of Plain descent*

Clinic *for* Special Children®

The Testing Process

Step 1

- Complete and sign a consent and requisition form.
- Arrange to have your blood drawn at the Clinic for Special Children or through your healthcare provider.

Step 2

- The Clinic for Special Children will conduct the testing at our lab. Call us for current turnaround time. Testing can be prioritized for couples who are expecting a baby, so let us know if this is the case.

Step 3

- The Clinic for Special Children staff or your provider ordering the test will notify you via mail or by phone of your results and explain what they mean for you and your family. We will invoice you for testing when results are complete.
- If needed, the Clinic for Special Children or your provider will discuss recommended next steps for any further testing, treatment, or preventative plans.

How does this help my children?

Some couples learn that they're carriers for genetic conditions only after the birth of an affected child. In some cases, the baby is diagnosed after medical concerns become apparent, undergoing a host of invasive and expensive tests and creating worry and stress for the whole family.

Expanded carrier testing, like the PIP, can identify if you're at risk of having an affected baby before the baby is born. If a targeted treatment exists for a condition, like GA-1 or MSUD, the baby can then be treated before they show symptoms. If both parents are not carriers for a shared condition, the PIP can also provide reassurance that a couple's children are not likely to be affected by the conditions included on the panel. Please note that a few conditions found in the Plain community are not included on the PIP and require additional testing. These include Fragile X, myotonic dystrophy, and Friedreich ataxia. Let your provider know if you have any family history for these conditions.

If you or your provider are interested in ordering testing through the Plain Insight Panel™, or have any questions about genetic testing, please contact the Clinic for Special Children *(see contact information below)*.

Clinic *for* Special Children®

20 Community Lane
Gordonville, PA 17529

tel (717) 687-9407
fax (717) 687-9237

ClinicforSpecialChildren.org