

Propionic Acidemia (PA) Methylmalonic Acidemia (MMA) Cobalamin (vitamin B12) disorders

(Newborn screening result: Elevated C3
with or without elevated C3/C2 ratio)

Fact Sheet for Parents

All babies born in North Carolina are screened at birth for certain health problems that can be treated if caught early.

Your baby's screening result showed a higher-than-expected level of a substance called C3. This can be a sign of a rare genetic condition that affects how the body processes proteins, fats, or vitamin B12. These conditions include propionic acidemia (PA), methylmalonic acidemia (MMA), and cobalamin (vitamin B12) disorders. In some cases, this result can also mean you or your baby is not getting enough vitamin B12.

A screening result is not a final diagnosis. Your baby will be referred to a specialist for more testing to confirm what is causing the high C3 level.

What are PA, MMA, and cobalamin disorders?

PA, MMA and cobalamin disorders are rare genetic conditions that affect how the body processes certain nutrients from food. In MMA and PA, the body has difficulty breaking down some proteins and fats. In cobalamin disorders, the body cannot properly use vitamin B12. As a result, harmful substances can build up in the blood and tissues, which may lead to health problems if not treated. With early diagnosis, regular medical care, and treatment, many people with these conditions live healthy, active lives.

Infants with confirmed diagnoses may show health concerns as newborns.

What happens next?

A screening result is not a final diagnosis. Your baby will be referred to a genetics specialist for additional testing. It is important to complete this testing as soon as possible, because these conditions can be managed best when treatment starts early.

What are the signs and symptoms of PA, MMA, and cobalamin disorders?

Symptoms may not be visible at birth. Common symptoms in the first months of life in those with untreated diagnoses are:

- Poor weight gain or growth
- Weak/low muscle tone
- Vomiting
- Dehydration
- Feeding problems
- Low energy (lethargy)
- Pale appearance
- Eye problems (maculopathy/retinal degeneration)
- Seizures

If your baby has any of these symptoms, contact your baby's provider. If your baby's provider is not available, take your baby to the emergency department immediately.

How are PA, MMA, and cobalamin disorders treated?

Treatment differs depending on the specific diagnosis. A baby with PA, MMA, or a cobalamin disorder may need to follow a lifelong protein-restricted diet, meaning they have to limit how much protein they eat. Most individuals with one of these conditions may also take a special formula to make sure they get enough essential proteins and nutrients to grow and develop. Individuals may need vitamin B12 supplements, carnitine supplements or folic acid. Your baby's doctor may recommend additional medications as your child develops and grows into adulthood.

Where do I go for more information?

Use your phone's camera to scan the QR codes below.

PA:



[MedlinePlus](#)



[NBSIC](#)



[Organic Acidemia Association](#)

MMA:



[MedlinePlus](#)



[NBSIC](#)



[Organic Acidemia Association](#)

Cobalamin disorders:



[MedlinePlus](#)



[NBSIC](#)



[Organic Acidemia Association](#)



NC DEPARTMENT OF
HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human
Services Division of Public Health

www.ncdhhs.gov

<https://slph.dph.ncdhhs.gov/newborn/default.asp>

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This fact sheet was supported by Health Resources and Services Administration (HRSA), Department of Health and Human Services (HHS) as part of a financial assistance award totaling \$344,348, with 100 percent funded by HRSA/HHS. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA/HHS or the U.S. Government.