Congenital Central Hypoventilation Syndrome: What You Need to Know

Congenital central hypoventilation syndrome (CCHS) is a rare, but serious disorder in which your baby does not breathe enough while asleep, and in some cases, while he is awake. In this handout, you will learn about CCHS and its symptoms. You will also learn how doctors test for and treat CCHS.

WHAT IS CONGENITAL CENTRAL HYPOVENTILATION SYNDROME?

Congenital central hypoventilation syndrome (CCHS) is a rare, but serious condition in which your baby does not breathe enough while he is asleep. In some cases, babies also do not breathe enough while they are awake. Not breathing enough can affect your baby’s brain and development.

WHAT CAUSES CCHS?

CCHS is caused by a mutation (change) in the PHOX2B gene. In most cases, the change happens at random. The change is not anyone’s fault.

WHAT ARE THE SYMPTOMS OF CCHS?

Symptoms of CCHS can include:

- Reduced or shallow breathing, especially during sleep
- Blue color around the lips or in the skin, especially during sleep
- Low body temperature
- Sweating more than usual
- Pupils (hole inside the colored part of the eye) that do not shrink when exposed to light
- Problems with the digestive tract (mainly the large intestine, such as with conditions like Hirschsprung’s disease)

HOW DO DOCTORS TEST FOR CCHS?

Doctors can use the following tests to test for CCHS:

- Sleep study to check your baby’s breathing while he sleeps
- Nerve, heart and lung tests
- Genetic testing to check for the PHOX2B gene

HOW DO DOCTORS TREAT CCHS?

It is important to diagnose and treat CCHS as early as possible. Starting treatment early can help your baby’s brain and body develop properly. The care team will talk with you about which treatments are right for your baby.

The most common treatment for CCHS is a ventilator (breathing machine). Some babies need a ventilator only while sleeping. Others need it more often. Other treatments for CCHS can include:

- Not taking part in activities that require long periods of holding your breath. This can include swimming, breath-holding contests or other activities.
- Staying away from drugs or alcohol when your baby is older.
- Appointments with specialists (doctors who treat specific conditions or parts of the body)
- In rare cases, a pacemaker (device inside the body that helps the heart pump properly) to keep blood flowing well throughout the body

WILL MY BABY’S CCHS BE CURED?

There is no cure for CCHS. It is a lifelong condition. With early diagnosis and treatment, the care team can help your baby will grow and develop normally.