Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 1)

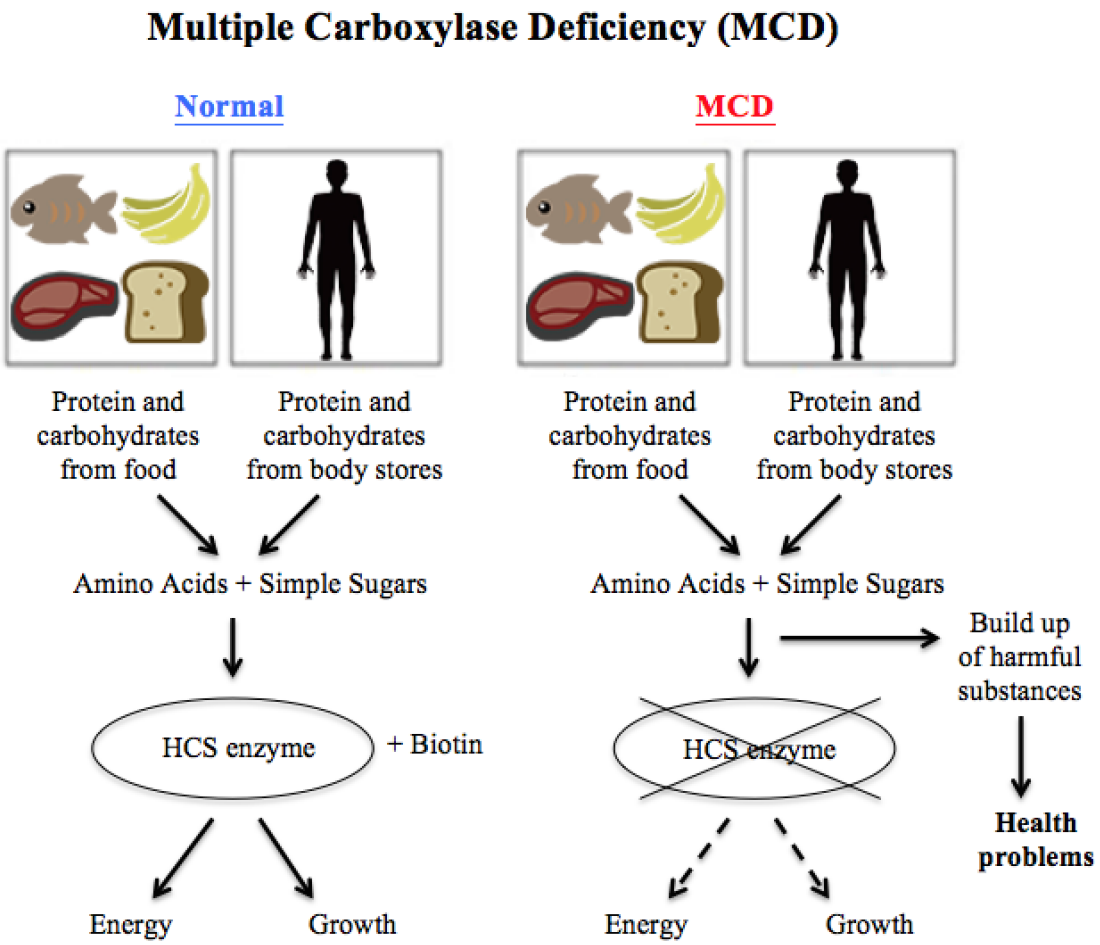
Multiple Carboxylase Deficiency

For general queries on Newborn Screening Programme for Inborn Errors of Metabolism, please call: ☎ 5741 4280 (Department of Clinical Genetics, Hospital Authority)

醫院管理局
HOSPITAL AUTHORITY

What is Multiple Carboxylase Deficiency (MCD)?

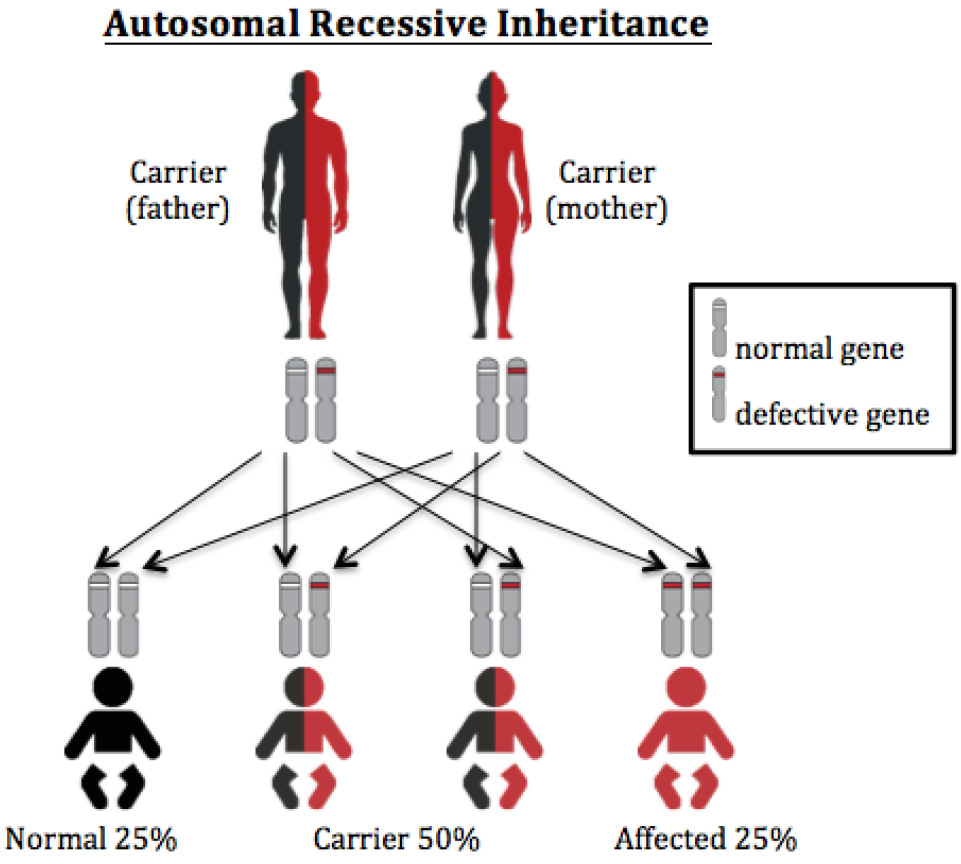
Multiple carboxylase deficiency is an inherited condition where the body is unable to use and recycle the vitamin biotin due to the deficiency of an enzyme called "holocarboxylase synthetase" (HCS). The body requires biotin to help process carbohydrates, proteins and fats. When this occurs, harmful substances can build up in the body and cause serious health problems.



How is Multiple Carboxylase Deficiency inherited?

Everybody has two copies of genes, one from each parent, which tell the body how to make specific enzymes.

Multiple carboxylase deficiency is an autosomal recessive disease. Only when babies inherit two faulty copies of the gene for multiple carboxylase deficiency from parents, the enzyme made does not work properly or is not even made at all.



What may happen if your baby has Multiple Carboxylase Deficiency?

In severe multiple carboxylase deficiency, symptoms may start as early as from a week to a few months after birth.

Some children with severe multiple carboxylase deficiency can experience metabolic crisis. These crisis are usually triggered when the body is under stress, such as when there is an infection or when there are prolonged periods without food.

Possible Signs and Symptoms of severe multiple carboxylase deficiency

Early signs include:

- Weak muscle tone
- Skin rashes
- Hair loss
- Hearing and vision loss
- Developmental delay

Some children may experience severe symptoms which include:

- Poor appetite and vomiting
- Extreme sleepiness
- Rapid breathing
- Seizures
- Coma

What is the treatment for Multiple Carboxylase Deficiency?

Babies with multiple carboxylase deficiency benefit significantly from early treatment and can have healthy and active lives.

Taking biotin supplement can prevent symptoms from occurring. This is a life-long treatment, and usually no dietary restrictions are required.

It is important that babies with severe multiple carboxylase deficiency be fed regularly and do not go for long periods without eating.

It is also important to discuss and design a possible plan with your doctor and dietician beforehand, in order to provide extra sugary foods during illness or other times when your child is not feeding well to prevent a metabolic crisis.

When should I seek immediate help? What should I do?

If you are worried that your baby is ill, it is important to follow medical advice. Bring your baby to your local accident and emergency department immediately. Take any information that you have been given about multiple carboxylase deficiency, including this pamphlet, to the hospital with you.