



Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 28)

Mitochondrial Trifunctional Protein deficiency (MTPD)

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



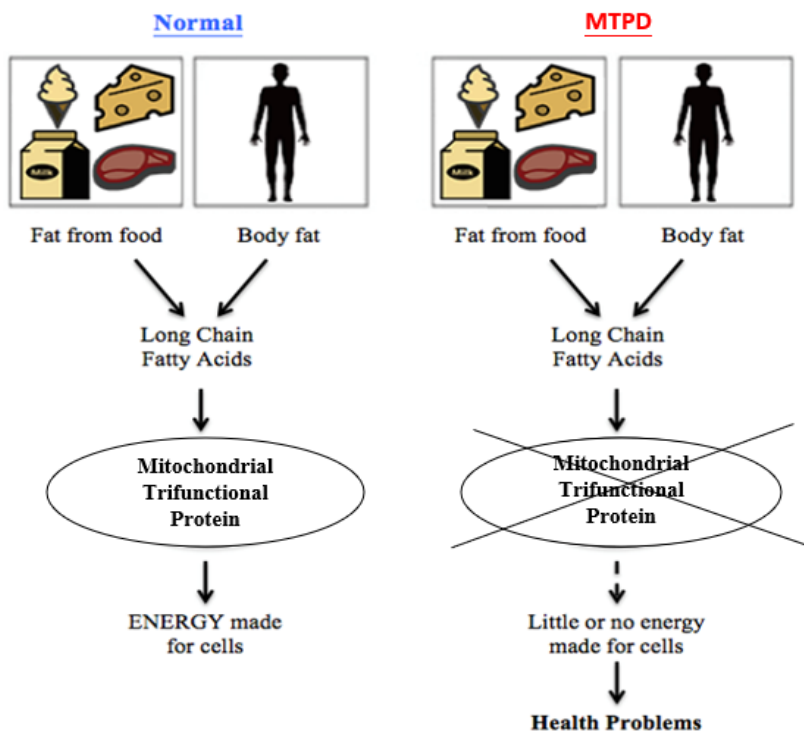
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What is Mitochondrial Trifunctional Protein deficiency (MTPD)?

MTPD is an inherited fatty acid oxidation disorder caused by deficiency or ineffective functioning of three enzymes called “long-chain enoyl-CoA hydratase, long-chain 3-hydroxyacyl-CoA dehydrogenase and long-chain 3-ketoacyl-CoA thiolase” (MTP). Without these enzymes, certain fats called “long-chain fatty acids” cannot be broken down properly.

Fatty acids are a major source of energy for the heart and muscles. During periods of fasting, fatty acids are also an important energy source for the liver and other tissues. When long-chain fatty acids are not metabolized properly, they cannot be converted to energy resulting in low blood sugar level (hypoglycaemia) and lethargy. At the same time, other toxic metabolites may also build up inside cells and damage multiple organs including the liver, heart and muscles.

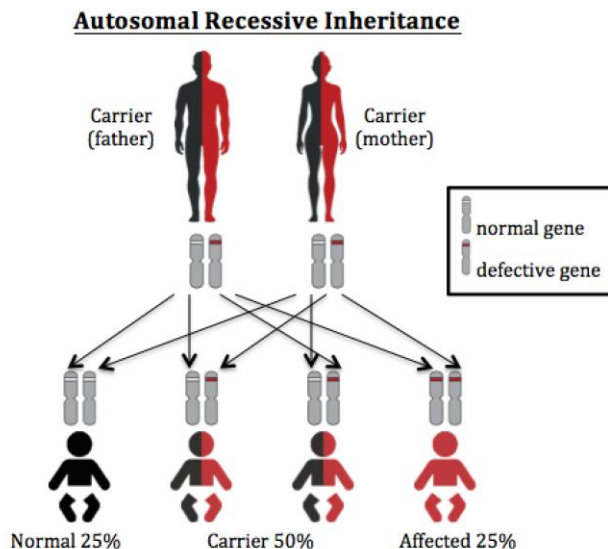
Mitochondrial Trifunctional Protein deficiency (MTPD)



How is MTPD inherited?

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

Mitochondrial Trifunctional Protein deficiency is an autosomal recessive disease. Only when babies inherit two faulty copies of the gene for mitochondrial trifunctional protein deficiency from parents, the enzyme made does not work properly or is not even made at all.



What may happen if your baby has MTPD?

MTPD can be very variable in presentation causing milder symptoms in some babies and more serious problems in others. Severely affected babies develop symptoms in the neonatal to infancy period and present in metabolic crisis. Metabolic crisis is a period of time when a metabolic disorder makes the baby seriously ill.

Mildly affected/late-onset type babies may develop symptoms in childhood or adolescence.

Possible Signs and Symptoms of MTPD

- ✚ Extreme sleepiness
- ✚ Behavioral changes / irritability
- ✚ Poor appetite / nausea / vomiting / diarrhea
- ✚ Muscle weakness
- ✚ Low blood sugar level (hypoglycaemia)

Other problems that some MTPD patients may have include:

- ✚ High levels of ammonia in the blood
- ✚ Enlarged liver and other liver problems
- ✚ Heart problems including enlarged heart and rhythm problem
- ✚ Breathing problems
- ✚ Muscle cramps, weakness, sometimes muscle breakdown after vigorous exercise
- ✚ Visual problems
- ✚ Nerve problems that may cause pain, numbness, tingling in different parts of the body

What is the treatment for MTPD?

Metabolic paediatricians and dieticians will work together to give expert advice and care to the babies with MTPD. When necessary, treatment is usually needed throughout the life. The goal of treatment is to prevent metabolic crisis and accumulation of toxic metabolites.

It is important that babies with MTPD be fed regularly and do not go for long periods without eating.

Treatment for MTPD usually includes

- a special diet with low-fat, high carbohydrate and MCT (medium chain triglyceride) supplement
- medication like L-carnitine may be advised for some patients but not the others. When indicated, L-carnitine is a safe and natural substance that helps body cells make energy. It also helps the body get rid of harmful wastes.

It is also very important is to discuss and design a care plan with your doctor and dietician beforehand, to let you know how to care for and provide extra sugary foods during illness or when your baby 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 MTPD, including this pamphlet, to the hospital with you.