Barth Syndrome

Barth syndrome is a rare *congenital* disorder. This means it is present since birth. This metabolic and neuromuscular disorder only affects boys. Barth syndrome affects at least 50 families worldwide. But there is evidence that it is under diagnosed.

CAUSES

- It is passed from mother to son through the sex-linked chromosome. This is the X chromosome.
- On average 50 percent of children born to a carrier mother will inherit the defective gene. But only boys will have symptoms.
- All daughters born to an affected male will be carriers.

SYMPTOMS

Problems (*symptoms*) affect many systems of the body. They may include:

- Changes to metabolism.
- Movement (*motor*) delays.
- Reduced muscle tone (*hypotonia*).
- Delayed growth.
- Longstanding (*chronic*) fatigue.
- Weakened immune system.
- Boys with the disorder also have fewer white blood cells. This is a condition called neutropenia. It may lead to an increased risk for serious germ (*bacterial*) infections.
- Lack of stamina.
- Low blood sugar (*hypoglycemia*).
- Mouth ulcers.
- Diarrhea.
- Cardiomyopathy. This would lead to a poorly functioning heart.
- Varying degrees of physical and learning disability.

TREATMENT

THERE IS NO SPECIFIC TREATMENT FOR THIS DISORDER.

- Bacterial infections caused by neutropenia can be effectively treated with medications which kill germs (*antibiotics*). The drug GCSF *(granulocyte colony stimulating factor*) can help fight infection. It stimulates white cell production by the bone marrow.
- Medicines may be prescribed to control heart problems.
- The dietary supplement carnitine has aided some children with Barth Syndrome, but in others it has caused increasing muscle weakness and has even precipitated heart failure.
- Only careful dietary monitoring directed by a physician or nutritionist familiar with the disorder can ensure proper caloric and nutritional intake.

Early and accurate diagnosis is key to prolonged survival for boys born with this disorder. Severe infections and heart failure are common causes of death in affected children.

RESEARCH BEING DONE

The NINDS supports research on genetic disorders such as Barth syndrome. It includes basic research on mitochondrial dysfunction and investigations of other inborn errors of metabolism. The 1996 discovery of the Barth gene, called G4.5 or TAZ1, is helping scientists and physicians better understand the metabolic and biochemical abnormalities seen in the disease. They are learning how genes cause:

- Heart disease.
- Muscle weakness.
- Other problems in the body.

The ultimate goal of this research is to find ways to prevent, treat, and cure these disorders.

Research is continually being done to learn more about this problem.

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