MSUD Family Support Group Contacts:

Executive Director
Denise Kolivoski, MBA
Erie, PA
814-580-8449
Execdirector@msud-support.org

President
Sandy Bulcher, BSN
Columbus, OH
740-972-5619
sandybulcher@gmail.com

Research Lead
Karen Dolins, EdD, RDN
Scarsdale, NY
914-391-2982
research@msud-support.org

Treasurer
Dave Bulcher
Columbus, OH
740-972-5618
davebulcher@gmail.com

We rely on donations to fund our newsletters, symposiums, and research projects to name a few. Please consider donating so that we can continue our work. See the MSUD Family Support Group website www.msud-support.org to donate to our Operating or Research Fund. We sincerely appreciate your donations!

For a complete list of support group contacts including our clinical and scientific advisors see the MSUD Family Support Group website.

MSUD

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The **Maple Syrup Urine Disease Family Support Group** is dedicated to:

- Connecting and engaging MSUD families and professionals
- Progressing research to improve the lives of those with MSUD
- Advocating to meet the needs of our community

What is MSUD?

www.msud-support.org

Maple syrup urine disease (MSUD) is an inherited disorder characterized by impaired protein metabolism, specifically the branched chain amino acids (BCAAs) leucine, isoleucine, and valine. The condition gets its name from the distinctive sweet odor of urine and earwax in affected infants. If untreated, MSUD can lead to developmental delays, seizures, coma, and death.

MSUD is a rare disease, affecting an estimated 1 in 185,000 infants worldwide. While pan ethnic and found world-wide, the disorder occurs much more frequently in certain ethnic groups, such as the Old Order Mennonite population with an estimated incidence of about 1 in 380 newborns, and the Ashkenazi Jewish population with an estimated incidence of 1 in 26,000 newborns.

MSUD was first described in 1954 and identified as an error in branched chain amino acid metabolism in 1957. The life-saving metabolic formula was developed in the 1960's, allowing survival for the first time. The formula provides all essential nutrients with the exception of the BCAAs leucine, isoleucine, and valine. These must be added to the diet in the precise amounts needed for normal growth and development. A strict diet and careful medical monitoring is essential throughout the lifespan of the affected individual.

Types & Symptoms

Maple Syrup Urine Disease includes both classic and variant types. The symptoms of classic MSUD are usually evident within the first week of life and include poor feeding, irritability, and the characteristic odor of maple syrup in the urine and earwax. Within days, infants may lose their sucking reflex and grow listless, develop a high-pitched cry, and become limp with episodes of rigidity. Without diagnosis and treatment, symptoms progress rapidly to seizures, coma, and death.

Variant forms of MSUD are milder and often not diagnosed until illness or infection cause symptoms to emerge. In variant types, a developmental delay or failure to thrive may be the first symptoms noticed.

Illness or infection can cause a life-threatening metabolic crisis at any age.

Diagnosing MSUD

All states in the US screen newborns for MSUD through newborn screening programs. Newborns should be tested 1-3 days after birth with results reported within 24 hours, as early diagnosis and treatment (within the first 10 days of life) reduces the risk of permanent damage and allows for normal development. In countries that do not screen for MSUD in the neonatal period, children with MSUD may become severely impaired or die before they are diagnosed.

Variant MSUD may be missed during newborn screening. Any child at risk or suspected of having MSUD should be tested promptly with a plasma amino acid panel and treated immediately if the results are positive or suspect.

An Inherited Disorder

MSUD is an autosomal recessive disorder. Each parent of a child with MSUD carries one abnormal gene for MSUD and one normal gene. Parents are called "carriers" and are not affected by the disorder as the normal gene is dominant (expressed). A child with MSUD has received an abnormal gene from each parent.

When both parents are carriers, there is a 1 in 4 chance with each pregnancy that the baby will receive an abnormal gene from each parent and develop MSUD; a 2 in 4 chance the baby will receive one abnormal and one normal gene, thus becoming a carrier of MSUD; and a 1 in 4 chance the baby will receive two normal genes. Persons with two normal genes cannot pass MSUD to their offspring.

Because a person with MSUD has two abnormal genes, a child born to that person will automatically be a carrier. However, the child will not have MSUD unless the other parent also has MSUD or is a carrier.

Treatment

Treatment of children with MSUD, which must be started as soon as possible, centers around a carefully controlled diet, the mainstay of which is a "medical food" or formula. The formula provides all essential nutrients except the 3 BCAAs for which there is impaired metabolism: leucine, isoleucine, and valine. These are added to the diet as prescribed by the treatment team based on regular monitoring of growth and blood amino acid levels. Food sources of protein are highly restricted. This regimen provides the child with adequate amino acids for growth and development while preventing high blood BCAA levels and symptoms. Careful monitoring of protein intake and close medical supervision is required for life.

Illness and stress, along with consuming too much protein from food, can raise levels of amino acids and their toxic breakdown products (keto acids) in the blood. Even a mild illness can become life-threatening if it prevents adequate intake of food and formula. Dietary changes are needed during these times and hospitalization may be necessary. Frequent blood tests allow clinicians to monitor amino acid levels.

Liver transplant is an optional treatment for MSUD as a donor liver has enough enzyme activity to allow the person to eat a normal diet and avoid metabolic crisis in most instances. After transplant, the person with MSUD still carries the gene for the disorder and will pass it on to their offspring. It is important to note that elevated branched chain amino acid levels have been observed in some transplanted individuals during illness.

MSUD Family Support Group

The MSUD Family Support Group is a non-profit 501 (c)(3) organization for those with MSUD and their families, health-care professionals, and others interested in MSUD.

Parents seeking information and support prompted the first MSUD Symposium for families and professionals in 1982. It was here that families discussed the need for an organized support group. Our support group has grown to include approximately 500 families and professionals worldwide. We publish a newsletter twice annually (one print, one electronic) and hold a biennial Symposium bringing together families, clinicians, and researchers.

In addition to connecting families and clinicians, our support group strives to promote research for improved treatments and a potential cure, raise awareness of MSUD, and advocate for legislation to meet the needs of our community.

To be added to our mailing list, please complete the information below. Donations support organizational costs that enable us to provide education and support to those affected by the disorder and their families. Your tax-deductible gift makes a difference!

MSUD FAMILY SUPPORT GROUP

☐ $25 ☐ $50 ☐ $100 ☐ $250 ☐ $Other _____

Name Professional Title (if applicable) Telephone ( )

Mailing Address City State Zip Country E-mail

Families: Spouse's Name MSUD Child's or Children's Name(s) Birth Date(s)

Make checks payable to: ***MSUD Family Support.*** *Mail completed forms to:*
Dave Bulcher, MSUD Family Support Group Treasurer • 4656 Winding Oak Dr, Delaware, OH 43015
Or go to the MSUD website www.msud-support.org to donate and sign up to receive mailings.