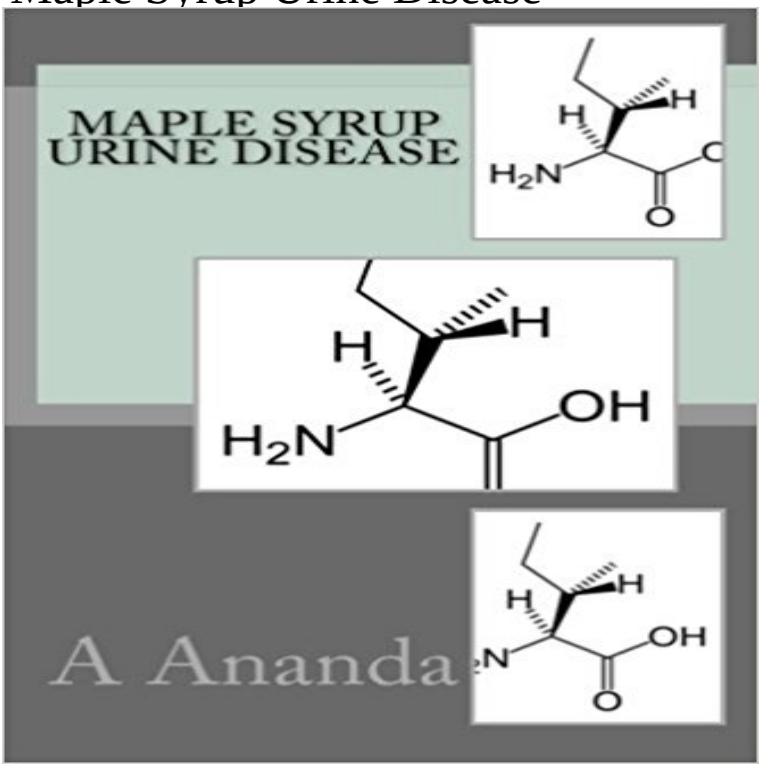
Maple Syrup Urine Disease



Maple Syrup Urine Disease (MSUD), also called branched-chain ketoaciduria, is rare, genetic disorder which is characterized by a deficiency of enzymes required to metabolize certain amino acids. These amino acids and their many metabolites abnormally accumulate in the cells and fluids of the body, causing symptoms including lethargy, irritability, convulsions, poor appetite and a maple syrup odor within the earwax, sweat and urine of affected people. If left untreated, seizures, coma and brain damage may occur. MSUD can be successfully managed by way of a specialized diet program.

OMIM Entry - # 615135 - MAPLE SYRUP URINE DISEASE, MILD Maple syrup urine disease (MSUD) is an aminoacidopathy secondary to an enzyme defect in the catabolic pathway of the branched-chain **Maple syrup urine disease type 2 Genetic and Rare Diseases** The American Chemical Society describes the lack of progress in understanding the pathology of MSUD. A collection of disease information resources and questions answered by our Genetic and Rare Diseases Information Specialists for Maple syrup urine disease. **OMIM Entry - # 615135 - MAPLE SYRUP URINE DISEASE, MILD** Maple syrup urine disease (MSUD), also called branched-chain ketoaciduria, is an autosomal recessive metabolic disorder affecting branched-chain amino **Maple Syrup Urine Disease Clinical Presentation: History, Physical** A number sign (#) is used with this entry because of evidence that a mild variant of maple syrup urine disease (MSUDMV) is caused by homozygous mutation in **Maple syrup urine disease** Maple syrup urine disease (MSUD) is a condition in which the body is unable to break down certain proteins. The condition is named for the sweet odor of the **Maple Syrup Urine Disease (MSUD) - Metabolic** Maple syrup urine disease (MSUD) is a rare genetic disorder characterized by deficiency of certain enzymes (branched-chain alpha-keto acid dehydrogenase **maple syrup urine disease - Genetics Home Reference Newborn screening information for maple syrup urine disease** A number sign (#) is used with this entry because of evidence that a mild variant of maple syrup urine disease (MSUDMV) is caused by homozygous mutation in **Maple Syrup Urine Disease - Symptoms, Diagnosis, Treatment of** MSUD Family support group. Dedicated to supporting families coping with Maple Syrup Urine Disease. **Orphanet: Intermediate maple syrup urine disease** Important It is possible that the main title of the report Maple Syrup Urine Disease is not the name you expected. Please check the synonyms listing to find the **Images for Maple Syrup Urine Disease** CLINICAL CHARACTERISTICS: Maple syrup urine disease (MSUD) is classified as classic or intermediate.

Twelve hours after birth, untreated neonates with **Orphanet: Classic maple syrup urine disease** Classic maple syrup urine disease (classic MSUD) is the most severe and probably common form of MSUD (see this term) characterized by a maple syrup odor **Maple Syrup Urine Disease: Background, Pathophysiology** Maple syrup urine disease (MSUD) is a metabolism disorder passed down through families in which the body cannot break down certain parts **Maple Syrup Urine Disease -** Maple syrup urine disease (MSUD) is a rare inherited disorder of branched-chain amino acid metabolism classically characterized by poor feeding, lethargy, **Maple Syrup Urine Disease (MSUD) - Healthline** Maple Syrup Urine Disease (MSUD) is a life-threatening rare genetic disorder present from birth. In MSUD, the body is unable to break down 3 amino acids **Maple syrup urine disease - NHS Choices** Maple syrup urine disease (MSUD) is an autosomal recessive disorder which can be caused by mutation in at least three genes. These genes encode the **Maple Syrup Urine Disease (MSUD) New England Consortium of** Introduction. Maple syrup urine disease (MSUD) is a rare but serious inherited condition. It means the body cant process certain amino acids **Maple syrup urine disease Genetic and Rare Diseases Information** Maple syrup urine disease (MSUD) is an aminoacidopathy secondary to an enzyme defect in the catabolic pathway of the branched-chain **Maple syrup urine disease: MedlinePlus Medical Encyclopedia Maple Syrup Urine Disease - WebMD** Maple syrup urine disease caused by mutation in the E1-alpha subunit gene is referred to as MSUD type IA that caused by a mutation in the E1-beta subunit **Maple Syrup Urine Disease - GeneReviews - NCBI Bookshelf** Drief description of MSUD and action pathway following detection of raised blood leucine level by newborn screening. **Maple Syrup Urine Disease Workup: Laboratory Studies** A collection of disease information resources and questions answered by our Genetic and Rare Diseases Information Specialists for Maple syrup urine disease **MSUD (maple syrup urine disease) - NEWBORN SCREENING** Maple syrup urine disease (MSUD) is classified as classic or intermediate. Twelve hours after birth, untreated neonates with classic MSUD **Orphanet: Maple syrup urine disease** Read our article and learn more on MedlinePlus: Maple syrup urine disease. **Maple syrup urine disease - Wikipedia** Maple syrup urine disease is an inherited disorder in which the body is unable to process certain protein building blocks (amino acids) properly **Maple Syrup Urine Disease - NORD (National Organization for Rare** Maple syrup urine disease (MSUD) is a metabolic disorder in which your body cant break down certain amino acids. Read more on how to **Maple Syrup Urine Disease. MSUD information and Causes Patient** Explains the inheritance mechanism for recessive conditions such as MSUD. **OMIM Entry - # 248600 - MAPLE SYRUP URINE DISEASE MSUD** Maple syrup urine disease (MSUD) is an aminoacidopathy secondary to an enzyme defect in the catabolic pathway of the branched-chain