Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 58350

Manuscript Type: CASE REPORT

Maple syrup urine disease: classic case reports of brain MRI findings and radiologic review

Li Y *et al*. MSUD MRI findings radiologic review

Abstract

BACKGROUND

Maple syrup urine disease (MSUD) is a rare autosomal-recessive disorder that affects branched-chain amino acid (BCAA) metabolism and is named after the distinctive sweet odor of affected infants' urine. This disease is characterized by the accumulation of BCAAs and corresponding branched-chain ketoacids of leucine, isoleucine, and valine in the plasma, urine, and cerebrospinal fluid. However, the mechanisms of MSUD-induced brain damage remain poorly defined. The accumulation of BCAAs in

31,700 Results Any time

Maple syrup urine disease | Radiology Reference Article ...

https://radiopaedia.org/articles/maple-syrup-urine-disease

Maple syrup urine disease (**MSUD**) is a very rare **metabolic disorder**. It is an inborn error of amino acid metabolism, which **classically** affects the brain tissue resulting in impairment or death if untreated. Epidemiology MSUD occurs in 1 in 185,0...

Maple syrup urine disease | Radiology Case | Radiopaedia.org

https://radiopaedia.org/cases/maple-syrup-urine-disease-8

Case Discussion. The features are characteristic of **maple syrup urine disease**. The patient had a family history of one sibling who had died on the 9th day of life with a clinical suspicion of **Maple syrup urine disease**.

Successful pregnancy in maple syrup urine disease: a case ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5948788

May 12, 2018 · Maple syrup urine disease (MSUD, OMIM 248600) is an autosomal recessive inborn error of branched-chain amino acid (BCAA) metabolism caused by deficiency of the branched-chain α-ketoacid dehydrogenase complex. Mutations in 3 of four genes that encode the catalytic subunits of the enzyme complex (E1, E2, E3) have been described.

Maple Syrup Urine Disease

Medical condition

Maple syrup urine disease (MSUD) is a rare but serious inherited condition.

NHS

Symptoms

Symptoms of MSUD usually appear within the first few days or weeks after birth. More general symptoms include:

- sweet-smelling urine and sweat
- poor feeding or loss of appetite
- weight loss

Babies with MSUD may also have episodes known as a metabolic...

Read more on NHS

Causes

国内版 国际版

Maple syrup urine disease: classic case reports of brain MRI finding

ALL IMAGES VIDEOS

58,500 Results Any time

Neuroradiological findings in maple syrup urine disease

https://www.ncbi.nlm.nih.gov/pmc/articles/pmid/23772241

Maple syrup urine disease is a rare inborn error of amino acid metabolism involving catabolic pathway of the branched-chain amino acids. This disease, if left untreated, may **cause damage to the brain and may even cause death**. These patients typically present with distinctive maple syrup odour of sweat and urine.

Imaging Findings in Maple Syrup Urine Disease: A Case ...

https://europepmc.org/article/MED/29899783

Jan 01, 2018 · I NTRODUCTION. **Maple syrup urine disease** (MSUD) is a rare autosomal-recessive disorder of branched-chain amino acid (BCAA) metabolism. It has an annual incidence of 1 in 180,000 live births worldwide.[1,2] Affected infants initially present with lethargy, irritability, feeding problems, and vomiting.If untreated, the **disease** progresses to cause seizures, coma, and eventually, death.[]

Author: Anjaneya S Kathait, Paulo Puac, Maurici... **Publish Year:** 2018

Case report: maple syrup urine disease with a novel DBT ...

https://bmcpediatr.biomedcentral.com/articles/10.1186/s12887-019-1880-1

Dec 13, 2019 · **Maple syrup urine disease** (MSUD) is a potentially life-threatening metabolic disorder caused by decreased activity of the branched-chain α-ketoacid dehydrogenase (BCKD) complex. Mutations in four genes (BCKDHA, BCKDHB, DLD and DBT) are associated with MSUD. Here, the presenting symptoms and clinical course of a **case** of MSUD with a novel DBT gene mutation are ...

Cited by: 5 **Author:** Wei Feng, Jinfu Jia, Heyang Guan, Qing Tian

Publish Year: 2019

Maple syrup urine disease: findings on CT and MR scans of ...

www.ajnr.org/content/11/6/1219

Imaging Findings in Maple Syrup Urine Disease: A Case Report

https://www.researchgate.net/publication/325478502...

Download Citation | **Imaging Findings** in **Maple Syrup Urine Disease**: A **Case** Report | **Maple syrup urine disease** (MSUD) is a rare autosomal-recessive …

Maple syrup urine disease: metabolic decompensation ...

https://europepmc.org/article/MED/8489211

Apr 01, 1993 · Metabolic decompensation of **maple syrup urine disease** in a 3.5-year-old boy was monitored by means of proton **magnetic resonance imaging** and spectroscopy. In the acute stag…

Cited by: 96 **Author:** S. R. Felber, W. Sperl, A. Chemelli, Ch. …

Publish Year: 1993

Maple syrup urine disease: Metabolic decompensation ...

https://www.onlinelibrary.wiley.com/doi/abs/10.1002/ana.410330412

Metabolic decompensation of **maple syrup urine disease** in a 3.5-year-old boy was monitored by means of proton **magnetic resonance imaging and** spectroscopy. In the acute stage, **imaging**…

Cited by: 96 **Author:** S. R. Felber, W. Sperl, A. Chemelli, Ch. …

Publish Year: 1993

Imaging findings of anaplastic astrocytoma in a child with ...

https://www.scholars.northwestern.edu/en/...

Here, we present the first **case** report of an anaplastic astrocytoma in a teenager with MSUD, with a discussion of **imaging findings** and the use of **magnetic resonance** spectroscopy (MRS) to hel…

Author: Jessie Aw-Zoretic, Jessie Aw-Zoreti… **Publish Year:** 2015

Wernicke-like encephalopathy during classic maple syrup ...

https://www.ncbi.nlm.nih.gov/pubmed/22350544

We describe a new neuroradiologic picture observed during metabolic decompensation in two **maple syrup urine disease** (MSUD) patients that resembles Wernicke encephalopathy (WE)….

Cited by: 8 **Author:** R. Manara, M. Del Rizzo, A. P. Burlina, A…

Publish Year: 2012