Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 58935

Manuscript Type: CASE REPORT

Neonatal isovaleric acidemia in China: A case report and literature review

Wu F *et al*. neonatal isovaleric acidemia

Abstract

BACKGROUND

Isovaleric acidemia (IVA) is a rare autosomal recessive inherited organic acidemia caused by a genetic deficiency of isovaleryl-CoA dehydrogenase (IVD). Its morbidity is low, but mortality is high. There is no effective cure for this disease. Early identification of IVA using clinical features can significantly slow disease progression and reduce mortality. Here we report a Chinese neonate with two mutations of IVD and share valuable information on this disease.

国内版 国际版

Neonatal isovaleric acidemia in China: A case report and literature r

ALL IMAGES VIDEOS

13,700 Results Any time

(PDF) Isovaleric Acidemia: A Rare Case of an Inborn Error ...

https://www.researchgate.net/publication/339600043...

PDF | **Isovaleric acidemia** (IVA) is an autosomal recessive disease of the leucine metabolism due to a deficiency of isovaleryl-CoA dehydrogenase (IVD).... | Find, read and cite all the research you ...

Eight novel mutations detected from eight Chinese patients ...

https://www.sciencedirect.com/science/article/pii/S0009898119320145

Nov 01, 2019 · 1. Introduction. **Isovaleric acidemia** (IVA, OMIM #243500) is a rare autosomal recessive disorder that is characterized by impaired leucine metabolism, and is caused by isovaleryl-CoA dehydrogenase (IVD) gene defect [].It is the first organic **acidemia** discovered in 1966 and is associated with significant morbidity and mortality [].The incidence of IVA varies among geographic regions, e.g., ...

Cited by: 1 **Author:** Yanhan Li, Ming Shen, Ying Jin, Yi Liu, Lulu ...

Publish Year: 2019

(PDF) Isovaleric Aciduria a Case Report of Two Patients

https://www.researchgate.net/publication/335745237...

Isovaleric aciduria (MIM 243500) is known as one of the major organic acidurias and is caused by a genetic deficiency of isovaleryl- CoA dehydrogenase (IVD: E.C.1.3.99.10) catalyzing the third ...

A Common Mutation Is Associated With a Mild, Potentially ...

https://pubmed.ncbi.nlm.nih.gov/15486829

Isovaleric acidemia: Therapeutic response to supplementation with glycine, l-carnitine, or both in combination and a 10-year follow-up **case** study. Chinen Y, Nakamura S, Tamashiro K, Sakamoto O, Tashiro K, Inokuchi T, Nakanishi K. Chinen Y, et al. Mol Genet Metab Rep. 2017 Mar 17;11:2-5. doi: 10.1016/j.ymgmr.2017.03.002. eCollection 2017 Jun ...

国内版 国际版

Neonatal isovaleric acidemia in China: A case report and literature r

ALL IMAGES VIDEOS

17,500 Results Any time

(PDF) Isovaleric Acidemia: A Rare Case of an Inborn Error ...

https://www.researchgate.net/publication/339600043...

Isovaleric acidemia (IVA) is an autosomal recessive disease of leucine metabolism due to deficiency of isovaleryl-CoA dehydrogenase (IVD). In this **case report** a five years old boy was admitted to ...

(PDF) Isovaleric Aciduria a Case Report of Two Patients

https://www.researchgate.net/publication/335745237...

Isovaleric aciduria (MIM 243500) is known as one of the major organic acidurias and is caused by a genetic deficiency of isovaleryl- CoA dehydrogenase (IVD: E.C.1.3.99.10) catalyzing the third ...

Eight novel mutations detected from eight Chinese patients ...

https://www.sciencedirect.com/science/article/pii/S0009898119320145

Nov 01, 2019 · 1. Introduction. **Isovaleric acidemia** (IVA, OMIM #243500) is a rare autosomal recessive disorder that is characterized by impaired leucine metabolism, and is caused by isovaleryl-CoA dehydrogenase (IVD) gene defect [].It is the first organic **acidemia** discovered in 1966 and is associated with significant morbidity and mortality [].The incidence of IVA varies among geographic regions, e.g., ...

Cited by: 1 **Author:** Yanhan Li, Ming Shen, Ying Jin, Yi Liu, Lulu ...

Publish Year: 2019

PEOPLE ALSO ASK

What is isovaleric acidemia (Iva)?

Does glycine reduce isovaleric acidemia?

21,200 Results Any time

(PDF) Isovaleric Acidemia: A Rare Case of an Inborn Error ...

https://www.researchgate.net/publication/339600043...

Isovaleric acidemia (**IVA**) is an **autosomal recessive disease** of the leucine metabolism due to a deficiency of **isovaleryl-CoA** dehydrogenase (**IVD**). We **report** the **case** of a six-month-old girl admitted...

Eight novel mutations detected from eight Chinese patients ...

https://www.sciencedirect.com/science/article/pii/S0009898119320145

Nov 01, 2019 · 1. Introduction. **Isovaleric acidemia** (IVA, OMIM #243500) is a rare autosomal recessive disorder that is characterized by impaired leucine metabolism, and is caused by isovaleryl-CoA dehydrogenase (IVD) gene defect [].It is the first organic **acidemia** discovered in 1966 and is associated with significant morbidity and mortality [].The incidence of IVA varies among geographic regions, e.g., ...

Cited by: 1 **Author:** Yanhan Li, Ming Shen, Ying Jin, Yi Liu, Lulu ...
Publish Year: 2019

Isovaleric acidemia: New aspects of genetic and phenotypic ...

https://onlinelibrary.wiley.com/doi/10.1002/ajmg.c.30089

Yasutsugu Chinen, Sadao Nakamura, Kunihito Tamashiro, Osamu Sakamoto, Kyoko Tashiro, Takahiro Inokuchi, Koichi Nakanishi, **Isovaleric acidemia**: Therapeutic response to supplementation with glycine, l -carnitine, or both in combination and a 10-year follow-up **case** study, Molecular Genetics and Metabolism **Reports**, 10.1016/j.ymgmr.2017.03.002, 11 ...

Cited by: 173 **Author:** Jerry Vockley, Regina Ensenauer
Publish Year: 2006

PEOPLE ALSO ASK

What is isovaleric acidemia (Iva)?

Does glycine reduce isovaleric acidemia?

What causes ivevaleric acidemia?