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Metabolic and Genetic Assessments Interpret an Unexplained .

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7,620 Results Any time

Guidelines for diagnosis and management of the cobalamin ...

https://onlinelibrary.wiley.com/doi/10.1007/s10545-016-9991-4

However nowadays, the **molecular genetic diagnosis** is the most advisable method provided the causal mutations in the index case and carrier status in the parents have been identified. **Molecular genetic testing** can be performed from chorionic villi or amniotic fluid samples (Morel et al 2005). Outcome: valid, timely laboratory diagnosis

Cited by: 30 **Author:** Martina Huemer, Daria Diodato, Bernd Sc...
Publish Year: 2017

[XLS] NICHD-NRNT_NCIt_Subsets 2014-01-13 - evs.nci.nih.gov

https://evs.nci.nih.gov/ftp1/NICHD/Neonatal... · Web view

A **cardiopulmonary disorder** characterized by systemic **arterial hypoxemia** secondary to **pulmonary hypertension** and extrapulmonary right to left shunting across the foramen ovale and ductus arteriosus. Neonatal Thrombocytopenia A condition characterized by a decrease in the number of platelets in the blood below established reference ranges in a newborn.

Pathophysiology, diagnosis, and treatment of methylmalonic ...

https://www.researchgate.net/publication/8417676_Pathophysiology_diagnosis_and...

Pathophysiology, diagnosis, **and** treatment of **methylmalonic** aciduria - Recent advances and new challenges Article · Literature Review in Pediatric Nephrology 19(10):1071-4 · November 2004 with 77 ...

Combined methylmalonic acidemia and homocystinuria, cblC ...

https://www.researchgate.net/publication/51485002_Combined_methylmalonic_acidemia_and...

Methylmalonic acidemia (MMA) is an autosomal-recessive **inborn metabolic disorder** that results from a **deficiency in methylmalonyl-coenzyme A mutase** or its cofactor, adenosylcobalamin.

AM Theft 2.0 Flashcards | Quizlet

https://quizlet.com/245006258/am-theft-20-flash-cards

Group 2 = most common, L heart disease (**pulmonary** venous **hypertension**, high capillary wedge pressure) - L heart failure due to systolic or diastolic dysfunction, valvular disease Group 3 = chronic lung disease - COPD, interstitial lung disease, obstructive sleep apnea, high altitude Group 4 = chronic thomboembolic PH - PH after PE Group 5 ...

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 52699

Manuscript Type: CASE REPORT

Metabolic and genetic assessments interpret an unexplained aggressive pulmonary hypertension induce by methylmalonic acidemia: A case report

Hong-Yu Liao, Xiao-Qing Shi, Yi-Fei Li

Abstract

BACKGROUND

Pulmonary hypertension (PH) causes significant morbidity and mortality in diverse

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Metabolic and genetic assessments interpret an unexplained a

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Three new cases of late-onset cblC defect and review of ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4255922

Reports on 58 cases (30 females, 22 males, 6 = no information) and the three new cases underlined the clinical heterogeneity of the disease. Time between first symptoms and diagnosis ranged from three months to more than 20 years. Haemolytic uraemic syndrome and **pulmonary hypertension** were main presenting symptoms in preschool children.

Cited by: 59 **Author:** Martina Huemer, Martina Huemer, Sabine...
Publish Year: 2014

Three new cases of late-onset cblC defect and review of ...

https://ojrd.biomedcentral.com/articles/10.1186/s13023-014-0161-1

Nov 15, 2014 · In general, inborn errors of metabolisms should be considered in **unexplained** medical cases at any age, especially in patients with multisystemic disease. More specifically, total homocysteine in plasma and **methylmalonic** acid in urine/plasma should be measured in **unexplained** neurologic, psychiatric, renal, haematologic and thromboembolic disease.

Cited by: 59 **Author:** Martina Huemer, Martina Huemer, Sabine...
Publish Year: 2014 **Author:** Martina Huemer

Combined methylmalonic acidemia and homocystinuria, cblC ...

https://www.researchgate.net/publication/51485002_Combined_methylmalonic_acidemia_and...

Combined **methylmalonic acidemia and** homocystinuria, cblC type. II. Complications, pathophysiology, and outcomes Article · Literature Review in Journal of Inherited **Metabolic** Disease 35(1):103-14 ...

(PDF) Combined Pulmonary Hypertension and Renal ...

https://www.researchgate.net/publication/256637153...

Combined **Pulmonary Hypertension and** Renal Thrombotic Microangiopathy in Cobalamin C Deficiency ... This study aims to improve our understanding of **methylmalonic acidemia** (MMA) complicated by ...

Will's study set Flashcards | Quizlet

https://quizlet.com/158794617/wills-study-set-flash-cards

Start studying Will's study set. Learn vocabulary, terms, and more with flashcards, games, and other study tools. Search. ... both an **assessment** for bilateral hearing loss and a neurologic examination. Sudden sensorineural hearing ... **pulmonary hypertension** C) atrial fibrillation D) angina pectoris E)

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Metabolic and genetic assessments interpret an unexplained a

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8,150 Results Any time

Three new cases of late-onset cblC defect and review of ...

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4255922

Reports on 58 cases (30 females, 22 males, 6 = no information) and the three new cases underlined the clinical heterogeneity of the disease. Time between first symptoms and diagnosis ranged from three months to more than 20 years. Haemolytic uraemic syndrome and **pulmonary hypertension** were main presenting symptoms in preschool children.

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Cited by: 59 **Author:** Martina Huemer, Martina Huemer, Sabine...
Publish Year: 2014 **Author:** Martina Huemer

Cobalamin C Defect Presenting With Isolated Pulmonary ...

https://www.researchgate.net/publication/237097415_Cobalamin_C_Defect_Presenting_With...

The concept of cardiorenal syndrome with **pulmonary hypertension** and TMA in MMACHC deficiency is supported by hemolytic anemia in an infant with cblC defect who died from cor pulmonale [49] and by ...

Methylmalonic aciduria: Newborn screening in mainland ...

https://www.researchgate.net/publication/235750381_Methylmalonic_aciduria_Newborn...

In 5 patients with **pulmonary hypertension**, 2 died, 2 recovered, and 1 **case** had mildly elevated **pulmonary** artery pressure. Seven patients underwent MMACHC gene testing, and 5 ...

2016 MCQs Flashcards | Quizlet

https://quizlet.com/243365217/2016-mcqs-flash-cards

• According to **case report** in clinical neuroscience "The vertebrobasilar system may play a role in TGA "**A case** of ... Other pharmacologic drugs occasionally used to **induce pulmonary** ... This results in **pulmonary hypertension**, signs of which include a narrowly split second heart sound, increased intensity of the **pulmonary** component of the ...