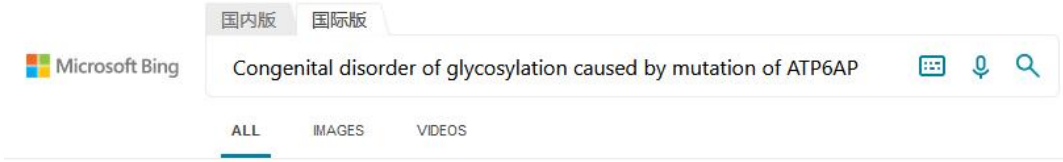

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Jul 17, 2020 · Feb 14th, 2020 - The **ATP6AP1 gene** encodes for ATPase H+ transporting protein. **ATP6AP1 gene mutations** are **associated with congenital disorders** of **glycosylation** (CDG) and can affect multiple organ system. Descriptions of postnatal phenotype include immunodeficiency, hepatopathy and cognitive impairment.

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https://www.sciencedirect.com/science/article/pii/S1590865821000979

May 01, 2021 · **Congenital disorders of glycosylation (CDG)** are multiorgan **diseases,** with variable degree of liver involvement in about 20% of types. Some **CDGs** have a predominant or isolated liver involvement (eg. MPI-CDG, CCDC115-CDG, TMEM199-CDG and ATP6AP1-CDG) and may present with ALF in …

Cited by: 1 **Author:** A. Di Giorgio, E. Bartolini, P.L. Calvo, M. Can…
Publish Year: 2021

Glyco25, XXV International Symposium on Glycoconjugates ...

https://link.springer.com/article/10.1007/s10719-019-09880-4

Jul 09, 2019 · **Congenital** defects **of glycosylation** (CDG) are inherited diseases due to hypoglycosylation of glycoproteins and glycolipids, and to defective biosynthesis of glycosylphosphatidylinositol anchors. The first clinical **report** appeared in 1980. Since then, the number of published CDG has shown an exponential expansion. Hundred thirty-five CDG are known.

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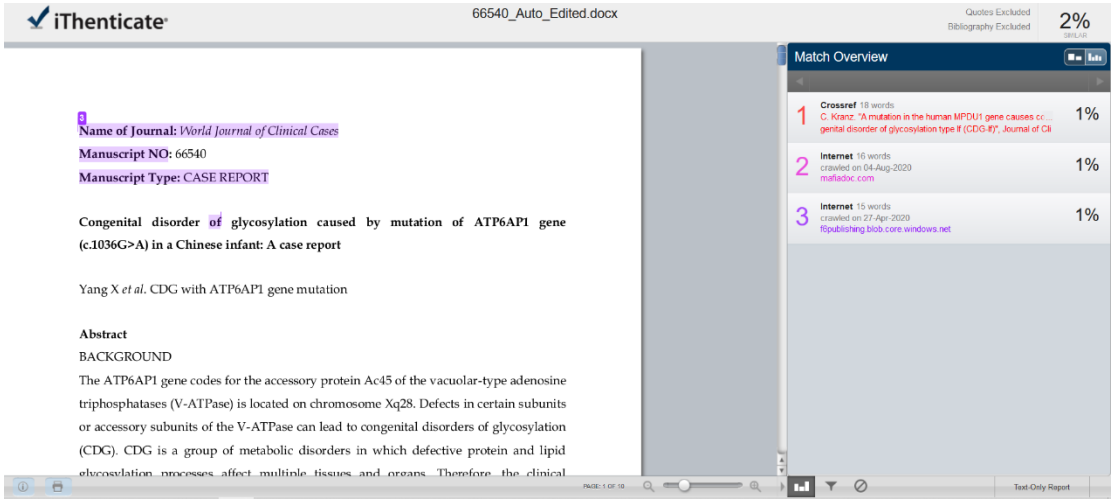

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 66540

Manuscript Type: CASE REPORT

Congenital disorder of glycosylation caused by mutation of ATP6AP1 gene (c.1036G>A) in a Chinese infant: A case report

Yang X *et al*. CDG with ATP6AP1 gene mutation

Abstract

BACKGROUND

The ATP6AP1 gene codes for the accessory protein Ac45 of the vacuolar-type adenosine triphosphatases (V-ATPase) is located on chromosome Xq28. Defects in certain subunits or accessory subunits of the V-ATPase can lead to congenital disorders of glycosylation (CDG). CDG is a group of metabolic disorders in which defective protein and lipid glycosylation processes affect multiple tissues and organs. Therefore, the clinical

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Jul 17, 2020 · Apr 15th, 2020 - **Congenital** disorders **of glycosylation** are a growing group of rare genetic disorders **caused** by deficient protein and lipid **glycosylation**. Here, we **report** the clinical, biochemical, and molecular features of seven patients from four families with GALNT2-**congenital disorder of glycosylation** (GALNT2-CDG), an O-linked **glycosylation** ...

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www.science.gov
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May 31, 2018 · Comprehensive PKD 1 and PKD 2 **Mutation** Analysis in Prenatal Autosomal Dominant Polycystic Kidney Disease. PubMed. AudrÃ©zet, Marie-Pierre; Corbiere, Christine; Lebbah, Said; Mor

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https://clinvarminer.genetics.utah.edu/.../gene/IQCB1
List of variants in **gene** IQCB1 studied for abdominal and pelvic region **disorder** Included ClinVar conditions (1028): 3 beta-Hydroxysteroid dehydrogenase deficiency

List of variants in gene TYR reported as pathogenic