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### First report of Gaucher disease in Montenegro: Genotype/phenotype ...

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作者: S Medenica - 2018

First report of Gaucher disease in Montenegro: Genotype/phenotype correlations. Sanja Medenica<sup>1</sup>, Snezana Vujosevic<sup>1,2</sup>, Vesko Vujicic<sup>3</sup>, Milena Dapcevic<sup>3</sup>, ...

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### Phenotype/genotype correlations in Gaucher disease type I: clinical ...

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作者: FY Choy - 2007 - 被引用次数: 45 - 相关文章

2006年12月29日 - ... disease among Chinese patients: review on genotype/phenotype correlation ... In this report, we review 29 cases of Gaucher disease from 23 ...

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[https://www.researchgate.net/scientific.../2045865850\\_Snezana\\_Vujosevic](https://www.researchgate.net/scientific.../2045865850_Snezana_Vujosevic) - [翻译此页](#)

Snezana Vujosevic's research while affiliated with University of Montenegro and ... First report of Gaucher disease in Montenegro: Genotype/phenotype correlations ... ketoacidosis, hypertriglyceridemia and acute pancreatitis- a case report.

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A Case Report: FGF23-Related Tumor-Induced Osteomalacia in a Patient .... First report of Gaucher disease in Montenegro: Genotype/phenotype correlations.

**Name of Journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 43481

**Manuscript type:** CASE REPORT

**Gaucher disease in Montenegro - genotype/phenotype correlations: Five cases report**

Vujosevic S *et al.* GD in Montenegro

Snezana Vujosevic, Sanja Medenica, Vesko Vujicic, Milena Dapcevic, Nikola Bakic,  
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### Gaucher's disease: report of 11 cases with review of ...

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Jan 07, 2015 · Clinical manifestations. In children with Gaucher disease, acute hip lesions can be misinterpreted as Legg-Calvé-Perthes disease, and avascular necrosis of the hips is a common complication in individuals of all ages. In our study, bone pain was present in 33.5% of cases with pathologic fractures in two cases.

Cited by: 2

Author: Laila Essabar, Toufik Meskini, Najat La...

Publish Year: 2015

### Gaucher disease: Gene frequencies and genotype/phenotype ...

[https://www.researchgate.net/publication/12834616\\_Gaucher\\_disease\\_Gene\\_frequencies\\_and...](https://www.researchgate.net/publication/12834616_Gaucher_disease_Gene_frequencies_and...)

The diversity of GBA gene mutations from patients with **Gaucher disease** represented in the ICGG **Gaucher** Registry database and in the literature provides the basis for initial **genotype/phenotype** ...

### Phenotype, diagnosis, and treatment of Gaucher's disease ...

[https://www.thelancet.com/journals/lancet/article/PIIS0140-6736\(08\)61522-6/fulltext](https://www.thelancet.com/journals/lancet/article/PIIS0140-6736(08)61522-6/fulltext)

**Gaucher's disease** continues to be a model for applications of molecular medicine to clinical delineation, **diagnosis**, and treatment. Analyses of several thousand affected individuals have broadened the range of the **pan-ethnic disease** variants, provided initial **genotype** and **phenotype correlations**, and established the effectiveness of enzyme therapy.

Cited by: 486

Author: Gregory A Grabowski

Publish Year: 2008

### Clinical and genetic characteristics of Gaucher disease ...

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3286762>

Feb 14, 2012 · Considering the high proportion of the neuronopathic type in Korean patients with **Gaucher disease**, new therapeutic strategies targeting the central nervous system are required. More specific **genotype-phenotype correlations** should be searched in a greater number of patients, which will help develop **genotype**-specific treatment modalities.

Cited by: 16

Author: Ju Young Lee, Beom Hee Lee, Gu Hwa...

Publish Year: 2012

### Gaucher disease among Chinese patients: Review on ...

<https://www.sciencedirect.com/science/article/pii/S1079979606005377>

In this report, we review 29 **cases** of **Gaucher disease** from 23 unrelated patients and 6 patients from 3 non-consanguineous families. Among these patients, 13 were diagnosed as Type 1, 10 as Type 2, and 6 as Type 3.

Cited by: 62

Author: Francis Y.M. Choy, Weimin Zhang, Hui...

Publish Year: 2007