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Basic Study

Wilson disease in lebanon and regional countries: Homozygosity and hepatic phenotype predominance

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Abstract

AIM

To determine the phenotypes and predominant disease-causing mutations in Lebanese Wilson disease patients, comparing it with regional non-European

Wilson disease in lebanon and regional countries: Homozygosity and hepatic phe

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... determine the predominant WD mutations in Lebanon and regional Arab countries, as well ... In Lebanon, 83% of patients were homozygous for the disease causing ... Pure hepatic phenotype was predominant and was found in 28% of patients. ... The number of studies on WD from neighboring Arab countries was small.

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Besides biological Genotypic-phenotypic profile of Wilson disease patients in and ... was performed on Lebanon: comparison with regional Arab countries liver lysate of ... the predominant WD mutations in Lebanon and activation of β- oxidation and ... Results: In Lebanon, 83% of patients were homozygous for the disease ...

Currently Clinical Views on Genetics of Wilson's Disease - NCBI - NIH
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4733722/
Wilson's disease, also named hepatolenticular degeneration, is an ... Gene Mutation, Genetic, Hepatolenticular Degeneration, Phenotype, Wilson's Disease .... homozygous for H1069Q mutation present more frequently with hepatic signs. .... Ferenci P. Regional distribution of mutations of the ATP7B gene in patients with ...

Wilson Disease in Lebanon and Regional Arab Countries: Predominance of Hep

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20 Apr 2017 ... Pure hepatic phenotype was predominant and was found in 28% of patients. ... The number of studies on WD from neighboring Arab countries was small. ... In order to improve the pickup rate of WD in Lebanon and the region ...

Wilson Disease - CAGS - Centre for Arab Genomic Studies
www.cags.org.ae/ctga/details.aspx?id=255
Countries with reported incidence ... Wilson disease (WD) is an autosomal recessive disorder, characterized by ... Egypt; Kuwait; Lebanon; Oman; Palestine; Saudi Arabia ... Pro1273Leu mutations in the ATP7B gene hinge region of exon 18. ... likely to have a hepatic phenotype, severe liver disease, a mixed phenotype, and ...

Wilson Disease - GeneReviews® - NCBI Bookshelf
https://www.ncbi.nlm.nih.gov/books/NBK1512/
29 Jul 2016 ... Wilson disease is a disorder of copper metabolism that can present with hepatic, ... Liver disease includes recurrent jaundice, simple acute self-limited .... A single pathogenic variant, a15-bp deletion in the 1-kb promoter region (c. ... The phenotypic spectrum has further expanded through molecular genetic ...

Wilson's disease: A review of what we have learned - NCBI
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4678372/

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Identification and analysis of mutations in the Wilson disease gene ...
https://www.ncbi.nlm.nih.gov/pubmed/9311736
Identification and analysis of mutations in the Wilson disease gene (ATP7B): population frequencies, genotype-phenotype correlation, and functional analyses ... accumulation of copper in the liver and subsequently in the brain and other organs ... and produces an in-frame, 39-bp insertion in mRNA of patients homozygous, ...