
ARCTIC HEALTH
An information portal to issues affecting the health and well-being
of our planet's northernmost inhabitants

A novel splicing mutation causes an undescribed type of analbuminemia.

https://arctichealth.org/en/permalink/ahliterature58568

Author: Monica Campagnoli
Antonio Rossi
Lars Palmqvist
Anders Flisberg
Aimon Niklasson
Lorenzo Minchotti
Monica Galliano

Author Affiliation: Department of Biochemistry, A. Castellani, Università di Pavia, Italy.

Source: Biochim Biophys Acta. 2002 Jan 2;1586(1):43-9

Date: Jan-2-2002

Language: English

Publication Type: Article

Keywords: Adult
Consensus Sequence
Female
Genotype
Heteroduplex Analysis
Humans
Infant, Newborn
Iraq - ethnology
Male
Mutation
Polymerase Chain Reaction
Polymorphism, Single-Stranded Conformational
RNA Splicing
Research Support, Non-U.S. Gov't
Serum Albumin - deficiency - genetics
Sweden

Abstract: Analbuminemia is a rare autosomal recessive disorder manifested by the absence or severe reduction of circulating serum albumin in homozygous subjects. In this report we describe a new molecular defect that caused the analbuminemic trait in a newborn of Iraqi origin. When the parents' DNA was analyzed, both subjects were found to be heterozygous for the same mutation found in the infant. All the 14 exon and flanking intron sequences of the albumin gene were amplified via PCR and screened for mutations by SSCP and heteroduplex analysis. A mutation in the DNA region encoding exon 1 and its flanking intron was revealed by the presence of a heteroduplex. The fragment, which was directly DNA sequenced, contains a previously unreported single nucleotide change, consisting in a G to A substitution at nucleotide 118 in the structural gene of the human protein. This mutation, involving the first base of intron 1, destroys the GT dinucleotide consensus sequence found at the 5' end of most intervening sequences and causes the defective pre-mRNA splicing responsible for the analbuminemic trait.

PubMed ID: 11781148 View in PubMed