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Pietro Maffei's research works | University-Hospital of ...<https://www.researchgate.net/scientific-contributions/Pietro-Maffei-39400618>

Tarekegn Geberhiwet Alström Syndrome (ALMS) is an ultra-rare multisystem genetic disorder caused by autosomal recessive variants in the ALMS1 gene, which is located on chromosome 2p13.

Endocrine manifestations related to inherited metabolic ...<https://orjrd.biomedcentral.com/articles/10.1186/1750-1172-7-11>

Jan 28, 2012 - Mutations of the ALMS1 gene cause dysfunction of the primary cilia, an organelle involved in cell sensing, as in another model of genetic obesity, the Bardet-Biedl syndrome. The higher frequency of diabetes in Alström syndrome is explained by the specific role of ALMS1 in β -cell function and/or peripheral insulin signaling pathways, in ...

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Alström syndrome is caused by mutations in ALMS1, a large gene comprised of 23 exons and coding for a protein of 4,169 amino acids.

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Zhang JJ et al. Alström syndrome with Graves' hyperthyroidism

Juan-Juan Zhang, Jun-Qi Wang, Man-Qing Sun, De Xu, Yuan Xiao, Wen-Li Lu, Zhi-Ya Dong

Abstract

BACKGROUND

Alström syndrome (AS, OMIM ID 203800) is a rare disease involving multiple organs in children and is mostly reported in non-Chinese patients. In the Chinese population, there are few reports on the clinical manifestations and pathogenesis of AS. This is the

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