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Accuracy of a Genetic Test for the Diagnosis of Hypolactasia in Chilean Children: Comparison With the Breath Test.

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Abstract

BACKGROUND:
Lactase nonpersistence (LNP) in humans is a genetically determined trait. This age-dependent decrease of lactase expression is most frequently caused by single nucleotide polymorphisms in the regulatory region of the lactase (LCT) gene. The homozygous LCT-13,910C/C genotype (rs 4988235) predominates in Caucasian adults with LNP, and is useful for its diagnosis in this population. The accuracy of this genetic test (GT) has not been completely established in children or in a Latin-American population.

OBJECTIVES:
The aim of the study was to determine diagnostic accuracy of GT for LNP in Chilean children using the lactose breath test (BT) as a reference, and to compare diagnostic yield in preschool- (<6 years) and in school-age (≥6 years) children.

METHODS:
Children referred for BT for diagnosis of lactose malabsorption to the Gastroenterology Laboratory at Clínica Alemana, Santiago, from October 2011 to March 2012 were invited to participate. After informed consent, symptom questionnaires, both historic and post lactose ingestion were completed. H2 and CH4 in expired air and -13,910 C>T single nucleotide polymorphism by polymerase chain reaction, restriction enzyme analysis, and/or Sanger sequencing were determined. GT accuracy was calculated compared to BT as reference method. Diagnostic yield of GT in preschool- and school-age children was compared.

RESULTS:
Lactose malabsorption was detected by BT in 42 of 60 children (70%). Genotype -13,910C/C was identified in 41 of 60 patients (68%). GT showed 80% sensitivity, 63% specificity, and 74% accuracy for LNP in the preschool population. In school-age children values were higher, 85%, 80%, and 84%, respectively.

CONCLUSIONS:
GT results were significantly concordant with BT results for hypolactasia detection in Chilean children, particularly in those of age 6 years and older.