Biochemical and Genetic Study for Newborn With Galactosemia That Caused by GALT Gene Mutation in Nineveh Province

Abstract

Iraq contains many of the diseases that have never been counted or examined, including diseases related to food, which has deteriorated in recent years, and has rapid and direct impact especially in children category, one of these diseases is galactosemia.

The aim of this study was to determine the possibility of diagnosing galacatosemia among Iraqi newborns, and to find the relationship between the presence of the disease and galactose concentration, galactitol and gal-1-phosphate level. Also to determine the genetic variation of galactose-1-phosphate uridyl transferase (GALT) and analysis of common mutation in GALT gene.

This study included taken blood from: 73 babies of both sex at age group (2-10) months admitted to the children's hospitals in Mosul City (Ibn alather and Alkhansa) depending on the clinical symptoms of disease, and 25 healthy babies regarded as control group, and 21 mothers at age group (20-35) years. Measuring the concentration of galactose in serum with Galactose-1-PhosphateuridylytransferaseGALT enzyme activity, galactitole concentration and galactose -1- phosphate by ELISA technique was done and DNA samples were analyzed by the polymerase chain reaction, followed by digestion with restriction endonuclease enzyme.

The result showed decrease GALT enzyme level in babies with galactosemia (21.7 ± 0.45 pg\ml) and in non-diagnosed children (79.8 ± 1.44 pg\ml) as compared with its level in healthy babies (160.33 ± 0.93 pg/ml), and the level of

enzyme in mother at (20.5 ± 1.92 pg\ml). Also the result showed significant decrease of galactos level in mothers (3.658 ± 0.213 pg\ml), in babies with galactosemia (18.3 ± 0.13 pg\ml) and non-diagnoses group (32.8 ± 2.65 pg\ml) compare its level with control group (73.5 ±10 pg\ml). The result revealed significant increase in Gal-1-P level in babies with galacatosemia (114.4 ± 8.4 pg\ml) and in non-diagnoses (52.8 ± 6.8 pg\ml) compare with healthy babies (25 ± 2.76 pg\ml) and in mothers a non-significant decrease was recorded (25 ± 1.3pg/ml) relative to healthy, Also there was an increase in galactitol concentration in patients (113.4 ± 4.8pg/ml) and in non-diagnosed (63.5 ± 2.7 pg/ml) compared with controls (27.9 ± 1.8pg/ml) and in mothers a non significant decrease was recorded (18 ± 1.72pg/ml) relative to healthy. Also the result showed decrease in β-carotene, ascorbic acid and selenium levels in babies with galactosemia compare it's with healthy babies and no change in α-tocopherol level in case study.

According to the molecular level the results showed that infected with galactosemia in study cases resulting from N314D mutation in GALT gene, while the results showed the Q188R is not found in GALT gene GALT in patients with galactosemiain Nineveh province, as well as the results did not show any genetic variation in 1721 site of GALT gene in patients with galactosemia, The results showed that the mutation R67C is present in all samples studied, so this mutation did not have any impact in the patients, mother and control group,and results also showed that four of the samples were carriers A330V mutation. The result did not show any genetic variation in first region that consist from first to fifth exon of GALT gene, while the results showed that thirteen cases have genetic variation in second region that consisting from sixth to ninth exon, in addition to these results there is no genetic variation in the third regain which consist from tenth and eleventh exon of GALT gene.