Galactosaemia in black South African children

Abstract:

Objective: To evaluate the clinical and biochemical features of all black children confirmed to have galactosaemia from the KwaZulu Natal Province of South Africa. Design: Prospective laboratory study. Subjects: These included all black children with the presenting clinical features suggestive of the diagnosis of galactosaemia. Setting: Department of Chemical Pathology, King Edward VIII Hospital, Durban, South Africa. Method: In each case, urine was screened for the presence of a reducing substance using urinary dipstick followed by thin layer chromatography to establish the presence of galactosaemia. The diagnosis of galactosaemia was then confirmed by analysis of galactose-1 phosphate uridyl transferase (GALT) activity in the erythrocytes using the established Beutler enzyme assay procedure. Age and sex-matched samples were used as controls for GALT activity. The presenting clinical features of each patient on admission were also recorded. Interventions: Patients confirmed to have galactosaemia were immediately placed on a galactose free diet. Results: The age distribution of affected individuals varied from six weeks to 27 months with 60% of the children being males. The most common presenting clinical features were jaundice in 77% of the patients, failure to thrive 62%, and cataracts 54%. Four patients had complete absence of GALT activity. Two infants who displayed acute toxicity symptoms and positive urine galactose, exhibited normal GALT activity. Conclusion: GALT deficiency is the most common form of galactosaemia in black children in the KwaZulu Natal region. Cases of galactokinase or epimerase enzyme deficiency appear to be present. Further investigation is required to establish the occurrence and prevalence of the latter in affected individuals in this region.