3. Outcome of Treated Galactosemia children in India

Dr. Anil B. Jalan

Objective: Screen high-risk neonates and infants for Galactosemia. Administer galactose free diet and follow-up for outcome**.**

Subject and method: This study included 371 high-risk neonates (lethargy, vomiting, jaundice, hypoglycemia, hepatomegaly, bleeding diathesis, and ascites). And two older children referred for hepatomegaly, ascites and jaundice. Out of 371 high-risk neonates, we diagnosed Galactosemia in 22, of which only 11 followed-up regularly. These 11 neonates along with 2 older children formed the study group. Screening for Galactosemia was performed by DBS – GALT enzyme analysis using BIO-RAD kit and total galactose by Quantase kit. Samples for all these subjects were taken for mutation analysis of the GALT gene, in order to identify the spectrum of disease causing mutations.

Results: Out of these 13 babies, 2 newborns and one older child (1Yr) expired soon after the diagnosis with severe bleeding diathesis and hepatic decompensation. Of these 13, there were 10 males and 3 females. 4 babies were born out of 3rd degree consanguineous marriage, and 9 had no history of consanguinity. Mean age of onset was 7.27 days, however the mean age referral was 28 days. Presenting symptoms were: jaundice (11/13), failure to thrive (11/13), hepatomegaly (10/13), hypoglycemia (9/13), lethargy (8/13), convulsions (7/13), ascites (6/13), bleeding diathesis (5/13) and cataracts (2/13). Mean GALT enzyme levels were (0.344 Units/Gm Hb) with a cut-off of 2.4 Units/GmHb: and mean total galactose levels were 105.95 mgm/dL (NR < 4.5). All of them had massive Galactosuria on TLC-Sugar. All these children were treated with galactose free diet, with supplementation of glucose, antibiotics and anti-convulsants whenever necessary. Barring 3 babies who expired, remaining 10 follow-up at our clinic. One baby was noticed to have cataract at subsequent visit. Microcephaly (HC<2 SD for the age) was noticed in 2 out of 10 children. None of the children so far has shown thyroidal dysfunction. The child with low IQ had poor compliance with diet.

Conclusion: Though mass screening is still not available in India for Galactosemia, early detection and early institution of therapy is possible. The outcome of properly treated children seems to be good except for short stature and at times microcephaly and abnormal EEG.