SUMMARY
AND
CONCLUSION
Beta thalassemia trait (BTT) is heterogenous state for the beta thalassemia gene. This condition is usually asymptomatic. These cases are detected when their blood samples are examined in a population survey, family study or investigations for refractory anemia is done. The diagnostic criterion for BTT is HbA2 ≥ 3.5%. The aim of the present work was to assess the diagnostic values of various parameters which are usually used in most of the laboratories in India for the diagnosis of BTT. The parameters undertaken were PCV, reticulocyte%, RBC count, indices like MCV, MCH, MCHC, discriminant factors, RBC morphology, osmotic fragility, Naked eye single tube red cell osmotic fragility test (NESTROFT), HbA2 estimation & HbF%. A comparison of HbA2 values in 2 methods from the same hemolysate was done. In the clinic group one hundred anemic cases and family members of 34 beta thalassemia ( homozygous & heterozygous ) were studied. Total individuals were 228. 87 cases were BTT ( HbA2 ≥ 3.5% ), 12 cases of thal major, 3 cases of thal intermedia and 5 cases of sickle thalassemia, 121 non-thalassemia. Blood sample of 201 individuals of Lohana community was examined for incidence of BTT. 27 cases were screening test ( NESTROFT ) positive & 31 were NESTROFT negative and included for further study. Out of 58 cases 18 were BTT and 40 non-thalassemia. All the cases (thalassemia & nonthalassemia) were grouped as without iron deficiency & with iron deficiency TS ≤ 16% or serum ferritin ≤ 10 μg/l. The parameters were studied and evaluated their diagnostic values.
Most of the carriers of BTT have varying mild degree of anemia. Observations in the present study indicate that the RBC count was slightly raised in BTT. There was no significant difference between the RBC count in BTT and that of the normal. In almost all cases of BTT morphological abnormality in RBC was marked. The abnormalities were microcytosis, hypochromia anisocytosis, target cells and schistocytes. Basophilic stippling was found in few cases. The value of MCV between BTT and control cases was highly significant. But there was no significant difference between MCV of BTT with and without ID. There was no significant difference in MCH of BTT and normal control cases, but significant difference in MCH was found in male & female BTT with ID cases. In respect of MCHC there was no significant difference between BTT cases and normal control cases. But significant difference MCHC value was found in male & female BTT with ID. Osmotic fragility was lower in BTT but significant difference was found between BTT and normal cases. Naked eye single tube red cell osmotic fragility test (NESTROFT) was highly sensitive for BTT but not specific test for BTT.

There was no significant difference in serum iron (S.I.) and total iron binding capacity (TIBC) between BTT & control cases. On the other hand the difference in transferrin
saturation (TS) was significant. In BTT cases with ID there was no difference in TS between male and female whereas in BTT without ID there was difference in TIBC. The mean serum iron in male BTT without ID was more than mean SI in female BTT without ID cases was significant. Transferrin saturation (TS) in male BTT without ID was more than female BTT without ID. The present work showed that HbA2 level is not influenced by iron deficiency in BTT cases. This is similar with other worker's observation. Iron status was different in different BTT cases. This might be due to different degree of ineffective erythropoiesis. HbA2 estimation by paper electrophoresis with elution and by microchromatography in BTT and nonthalassemia cases showed positive correlation coefficient (r) in both methods. There was no effect of iron deficiency in HbA2 level in BTT cases in both methods. HbA2 estimation by paper electrophoresis was more sensitive than HbA2 by microchromatography method. Specificity for both methods were high. HbA2 estimation by paper electrophoresis and elution can be recommended as it is more sensitive, economical and one can see different Hb bands in electrophoresis strips. Majority of BTT had either no elevation or slight increase in HbF%.

Discriminant factors were more sensitive to IDA cases than BTT. The coexisting disease which influence the red cell count invalidates both MCV, DP2, DP1 as diagnostic indices in iron deficiency anemia and BTT. In population survey iron deficiency anemia with low Hb discriminant factors may not be helpful.
The commonest parameters were Hb gm%, MCV, HbA2, RBC morphology, osmotic fragility & NESTROFT, which were sensitive to significant number of cases individually and in more percentage in combination. HbA2 $\geq$ 3.5% has been accepted as the diagnostic criterion for diagnosis of BTT.

For population survey, initially there should be screening test from the parameters which are highly sensitive and commonly used for diagnosis of BTT. Naked eye single tube red cell osmotic fragility test (NESTROFT) satisfies all the criteria. It is simple and highly sensitive.

The prevention of thalassemia major programme should be started with population screening for BTT and then confirmation by HbA2 estimation and other sophisticated test if required. For prospective marriagable spouses marriage should be discouraged if both are beta thalassemia carrier. In pregnant woman prenatal diagnosis should be done. Facilities should be available for prenatal diagnosis with DNA analysis from chorion villi in 1st trimester of pregnancy. If it is not possible in the 1st trimester of pregnancy, fetal blood may be collected in 18th week of gestation for globin chain synthesis/DNA analysis. Mothers carrying the homozygous beta thalassemia fetus should be motivated for termination of pregnancy.