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Comparative study of solubility and hemoglobin electrophoresis with high performance liquid chromatography (HPLC) for screening of hemoglobinopathies and thalassemia: Study from central India

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Abstract

Introduction: *High prevalence of hemoglobinopathies is seen in central India, screening and genetic counseling are essential for early detection and management.*

Aim: *The retrospective study was performed at Regional hemoglobinopathy detection and management centre (RHDMC) Nagpur, central India to find out relative frequencies of hemoglobinopathies and thalassemia present using solubility test, hemoglobin electrophoresis and high performance liquid chromatography (HPLC) as screening method and to compare results of HPLC with solubility and electrophoresis.*

Materials and Methods: *A total of 105,211 cases were screened for sickle cell disease (SCD) and sickle cell trait (SCT) by solubility test during the period of January 2003 to January 2014. Of these 105,211 samples, 60,000 samples which were solubility positive, with doubtful solubility and solubility negative but suspicious for hemoglobinopathy and thalassemia also the cases of anemia were studied by hemoglobin electrophoresis at alkaline pH 8.6. Of which 5,111 cases were further studied by HPLC and results of HPLC were compared with combined solubility and Hb electrophoresis.*

Results: *Of 105,211 cases screened for hemoglobinopathy by solubility and electrophoresis, 12,979 (12.33%) were having sickle cell trait (SCT) and 3,062 (2.91%) were of sickle cell disease (SCD). Of 5,111 (100%) HPLC study cases, total SCD and SCT were 3,132 (61.27%) followed by 315 (6.16%) of beta-thalassemia trait and 264 (5.16%) cases of compound heterozygous for HbS and betathalassemia. Hemoglobinopathies E and D alone and its combination with HbS or beta-thalassemia were also found. Rare cases of HbD Iran, HbJ variant and HbQ India, Hb Abruzzo and delta-beta thalassemia were detected. Combined solubility and hemoglobin electrophoresis was effective for diagnosis of SCD and SCT when compared to HPLC with good agreement between two test by kappa statistics, however for detection of beta-thalassemia trait and for compound heterozygous for HbS and beta-thalassemia false negatives cases were more, chi square test showed highly significant P value* < 0.01.

Conclusion: *Combined solubility and electrophoresis are simple and cost effective alternative to HPLC for screening large population with high prevalence of SCD when resources are limited but for beta-thalassemia screening HPLC is mandatory.*

Author Keywords

Hemoglobinopathy, Thalassemia, HPLC, Solubility, Hemoglobin Electrophoresis

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