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Evaluation Of High Performance Liquid Chromatography Pattern And Prevalence Of Thalassemia Trait In Port Harcourt, Nigeria

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INTRODUCTION

Sickle cell disease (SCD) is the most prevalent haemoglobinopathy in Nigeria which is among the nations that has the highest burden of SCD. Other forms of haemoglobinopathies such as Thalassaemia were hitherto thought to be rare in Nigeria and probably restricted to Mediterranean regions. However, with increasing availability and use of more sensitive haemoglobin screening methods such as High Performance Liquid Chromatography(HPLC) Machine, there is increasing evidence of the prevalence of thalassaemia disorders in Nigeria. The HPLC machine quantifies the regular haemoglobin variants and estimates haemoglobin A_2 and F levels. The HbF and HbA_2 levels combined with evaluation of red blood cell indices serves as a suitable laboratory screening tool for the suspicion, diagnosis and categorization of patients with thalassaemia and those with compound sickle cell disease such as co-inheritance of S and thalassemia genes.

Classical laboratory finding in thalassemia include low haematocrit in severe disease, low red cell indices (mean corpuscular haemoglobin (MCH), and mean corpuscular volume (MCV)). On haemoglobin electrophoresis, a high A_2 levels >3.5% is suggestive of a beta thalassemia. HbF levels is usually elevated (>1%). Combining these parameters, assessment of alpha and beta thalassemia can be entertained. Globin chain and genetic studies can be used to make definitive diagnosis.

There is a growing number of researches on the burden of thalassemia in Nigeria. The findings from these studies suggest the presence of thalassemia disorder in Nigeria, but there is a dearth of such studies in Port Harcourt City, Nigeria.

AIM

The aim of this retrospective assessment was to determine the prevalence of Thalassemia trait in this cohort of patients using our service using the HbA2 level as measured by High Performance Liquid Chromatography.

METHOD

This was a retrospective review of a three year records of adults and children who were investigated for anaemia and haemoglobinopathies at our haematology clinic. Their haemoglobin analysis and blood counts were estimated using the HPLC system by BioRad D10 and Sysmex XP300 respectively.

RESULTS

One hundred and twenty-nine patients were evaluated during the period, 89 of them had complete details of their haemoglobin patterns including HbA_2 and HbF, red cell indices (MCV, MCH) were included in the analysis. Data was analysed with SPSS version 23 and summarized using descriptive statistics.

The median (IQR) of the subjects was 12years (6 – 21). They included 54 (60.7%) males and 35 (39.3%) females. The haemoglobin phenotypes for the patients were SS in 54 (60.7%), AS in 10 (11.2%), S-βThal in 10 (11.2%), SC in 2 (2.2%) AA in 6 (6.7%), CC in 1(1.1%) and 6 (6.7%) were thought to have a thalassemia syndrome.

Majority of the patients, 77 (86.5%) had anaemia with PCV < 30%, MCH was <27g/pl in 54 (60.7%) and MCV was <76fl in29 (32.6%). Eighty four (94.4%) of them had HbF >1%. A high HbA_2 >3.5% was found in 36 (40.4%) of the patients.

Using MCV < 76fl and HbF >1% in those with a high HbA_2, 13 (14.6%) had possible □-Thal trait, while 15 (16.9%) had silent □ - thalassemia trait.

Thalassemia status	Criteria	n (%)
Possible β thal	Low MCH, High A_2	23 (25.8)
Silent α thal trait	Low MCH, Normal A_2	31 (34.8)
Silent β thal trait	Normal MCH, High A_2	13 (14.6)
Normal	Normal MCH, Normal A_2	22 (24.7)
Thalassemia status	n (%) (MCV <76, HbF>1%)	n (%) (MCV<76, HbF <1%)
Possible β thal	13 (14.6)	0 (0.0)
Silent α thal trait	15 (16.9)	1 (1.1)

CONCLUSIONS

Our findings show that 60.7% of patients evaluated for haemoglobinopathies in our laboratory had haemoglobin SS, 3.3% were haemoglobin SC and for the first time we demonstrated that 6.7% of the patients had Sb Thal in Port Harcourt. There is need for further family studies and molecular evaluation to confirm these findings.

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