Evaluation of Cardiac Status of Patients with Haemoglobinopathies by 2D Echocardiography with Specific Reference to Pulmonary Hypertension

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Abstract: _Introduction: Haemoglobinopathies are the disorders affecting the structure, function, or production of haemoglobin. Hereditary haemoglobin disorders including mainly beta thalassemia and sickle cell anaemia represents most common monogenic disorders in population.1Different forms may present as ineffective erythropoiesis, haemolytic anaemia, erythrocytosis, cyanosis, or vasoocclusive stigmata.2 Material and methods: The study comprised of all diagnosed cases of haemoglobinopathies admitted in Medicine Department tertiary care centre between December 2017 to October 2019. The researcher has adopted prospective cross sectional design to find correlation of pulmonary arterial hypertension (PAH) with patient of haemoglobinopathies. Conclusion: This study highlights importance of screening for pulmonary hypertension in hemoglobinopathies to detect development of pulmonary hypertension at the earliest._

Keywords: Haemoglobinopathies

1. Introduction

The spectrum of cardiovascular manifestations in haemoglobinopathies is wide and it includes ventricular dysfunction, pulmonary hypertension, pericarditis, myocarditis, arrhythmias, stroke and thromboembolic events.

Pulmonary hypertension is a spectrum of diseases involving the pulmonary vasculature and it is defined as an elevation in pulmonary arterial pressure (PAP) >22mmHg or an estimated systolic PAP >36mmHg.3 If left untreated the disease carries a high mortality rate, with the most common cause of death being right heart failure.

The emergence of pulmonary hypertension as a complication of the hereditary hemolyticanemias is a major example of potential global impact of pulmonary vascular disorders. There has been significant progress in understanding the pathophysiology of this chronic complication of hemolytic diseases, but there is still a major need for the development of novel therapies for this patient population that could only be achieved if we direct our efforts toward including the developing world in multinational clinical trials to meet these challenges.5

Screening for Pulmonary hypertension should be an essential component of haemoglobinopathy assessment and may be accomplished by transthoracic Doppler echocardiography. Doppler echocardiography is the most common and established screening tool for PH. It is widely available, cost effective and is part of the normal cardiovascular examination of haemoglobinopathy patients.4

Pulmonary arterial hypertension (PAH) is characterized by a sustained increase in pulmonary artery pressure and a progressive increase in pulmonary vascular resistance, leading to right ventricular insufficiency. Classically, pulmonary hypertension was divided into primary (idiopathic) and secondary forms. However, within the secondary PAH category, there are conditions that are similar to those of primary PAH, in terms of histopathological characteristics as well as response to treatment. The World Health Organization has periodically offered classifications of PAH, the current classification being the result of a consensus meeting held in 2003 in Venice, Italy (Chart1). Mean pulmonary artery pressure (MPAP), under physiological conditions and at sea level, is < 20 mmHg, and pulmonary artery systolic pressure (PASP) is < 30 mmHg. The definition of PAH (group 1 of the Venice Classification) is based on hemodynamic criteria: MPAP > 25 mmHg at rest or > 30 mmHg during exercise, with pulmonary capillary pressure or left atrial pressure < 15 mmHg and pulmonary vascular resistance > 3 mmHg. Currently there is an updated classification (Dana Point, USA, 2008) that includes idiopathic pulmonary arterial hypertension, formerly named primary, in Group 1, being: hereditary (BMPR2, ALKI, endoglin), induced by drugs/toxins, related to systemic-pulmonary artery shunt and to hypertension due to persistence of fetal pulmonary circulation pattern; introducing new hemodynamic parameters of values for pulmonary artery pressure, where: normal < 21 mm Hg; boundary between 21 and 25 mm Hg, and evidenced pulmonary hypertension > 25 mm Hg. The principal alterations seen in PAH are vasoconstriction, remodeling and in situ thrombosis. There is endothelial dysfunction, which leads to impaired production of vasodilators, such as NO and prostacyclin, and increased expression of vasoconstrictors and mitogens, such as endothelin-1. Idiopathic PAH is more common in women than in men (ratio, 1.7: 1), and the mean age at diagnosis is 36 years. In many cases, the diagnosis of PAH is delayed, since the symptoms are nonspecific and can be confused with those of other, more common diseases. Dyspnea is the initial symptom in 90% of patients. Less common symptoms include fatigue, chest pain, syncope, peripheral edema and palpitations. Various diseases, such as portal hypertension, haemoglobinopathies, collagen diseases and

HIV infection, are associated with PAH. Recent studies have suggested an association between PAH and thyroid diseases (hypothyroidism and hyperthyroidism). Since the Third World Conference on Pulmonary Hypertension, heldin 2003, thyroid diseases have been classified as diseases associated with PAH. However, although some pathogenic mechanisms have been proposed, the nature of this association has not yet been established[8]

A large body of literature supports the potential role of intravascular hemolysis and abnormal NO signaling in the pathogenesis of precapillary pulmonary hypertension in SCD. Intravascular hemolysis leads to the release to the plasma of cell-free hemoglobin, submicron red blood cell microparticles that contain hemoglobin and haem, and arginase-1. The emptying of these red blood cell contents into plasma has the potential to inhibit NO signaling and impair vascular endothelial function. Cell-free plasma hemoglobin rapidly reacts with NO to form nitrate and erythrocyte-derived arginase-1 limits the availability arginine, the obligate substrate of the NO synthases. Release of haem from the hemoglobin molecule in plasma leads to activation of TLR4 and inflammatory pathways. Hemolysis also drives platelet and hemostatic activation and is proposed to generate reactive oxygen species and activate vascular oxidases. Clinical and translational studies have found associations of markers of hemolysis, including cell-free hemoglobin and red blood cell microparticles, with endothelial dysfunction, increased estimated systolic pulmonary artery pressure, and right heart catheterization–documented pulmonary hypertension. A trial of the 5′-phosphodiesterase inhibitor, sildenafil, to increase downstream mediators of the NO signaling pathway such as soluble guanylate cyclase and cyclic GMP in SCD patients with pulmonary hypertension or elevated TRV was conducted. The trial was stopped early because of high painful crisis rate in the sildenafil arm, possibly related to effects of inhibition of 5′-phosophdiesterase on limb pain, myalgia, and back pain. Although this study did not show a reduction in mean pulmonary artery pressure or TRV in subjects receiving sildenafil at the time it was stopped, other recent publications continue to offer support for the "hemolytic vasculopathy" view of SCD[6-10].

SCD is a condition with a predilection for thrombotic complications including pulmonary embolism, and CTEPH is a major category of pulmonary hypertension. Therefore, consideration needs to be given to the possibility that chronic thromboembolism may contribute to pulmonary hypertension in SCD. In one series, scintigraphic evidence suggestive of CTEPH occurred in approximately 12% of SCD patients with pulmonary hypertension. The clinical presentation is similar to Group 1, pulmonary arterial hypertension, but mismatched segmental defects on ventilation/perfusion scanning is suggestive of the diagnosis of CTEPH, and pulmonary angiography is confirmatory of the diagnosis. The interpretation of ventilation/perfusion scans can be complex in the setting of preexisting lung abnormalities, and pulmonary angiography is an invasive procedure. Nevertheless, making the diagnosis of CTEPH is important because pulmonary endarterectomy can be curative for patients with proximal obstructive lesions, and the precise diagnosis can guide medical therapy required for

patients with distal lesions who do not qualify for a surgical approach. Pulmonary endarterectomy poses a challenge to SCD patients because of the tendency for increased sickling during cardiopulmonary bypass, hypothermia, and circulatory arrest associated with the procedure. It is wise to reduce hemoglobin S to ≈10% by exchange blood transfusion before the procedure, and with this approach a number of pulmonary endarterectomy procedures have been successfully performed in SCD patients with CTEPH. Hemolysis promotes platelet and hemostatic activation in the setting of SCD, and chronic inflammation is another potentially contributing factor to a hypercoagulable state. Furthermore, most hemoglobin SS patients have autosplenectomy, and splenectomy is a recognized risk factor for thrombosis and CTEPH. In keeping with these observations, autopsy studies suggest the possibility of a contribution of thromboembolism in some cases of SCD pulmonary hypertension. Microthrombotic and/or thromboembolic lesions are common findings at postmortem examination of patients with SCD. Acute or organizing thrombi in the pulmonary arteries, predominantly distal, was a common fining at autopsy in a series of 11 SCD patients with the diagnosis of pulmonary hypertension. Plexiform lesions were reported to be another common finding, but at least some of these lesions may have actually represented recanalyzed thrombi. A significant association was found between the postmortem diagnoses of pulmonary hypertension and thromboembolism in another autopsy series[11, 12].

In general, the tricuspid regurgitation velocity (TRV) measured during echocardiography in combination with estimated right atrial pressure is considered to be a valid estimate of systolic pulmonary artery pressure. The normal TRV is considered to be <2.5 m/sec, but elevation of TRV and estimated systolic pulmonary artery pressure does not reliably identify subjects with pulmonary hypertension defined as mean pulmonary arterial pressure ≥25 mm Hg. Rather, an elevated TRV can help identify subjects who should be examined definitively with right heart catheterization. Pulmonary hypertension is considered to be unlikely if the TRV is ≤2.8 m/sec and there are no other echocardiographic changes suggestive of pulmonary hypertension such as enlargement of right-sided chambers and right ventricular systolic dysfunction. The diagnosis is considered to be possible if the TRV is 2.9-3.4 m/sec and to be likely if it is >3.4 m/sec. Several studies have documented a positive correlation of TRV and echocardiography estimated systolic pulmonary artery pressure with systolic pulmonary pressure measured at right heart catheterization in patients with SCD. In these studies, the correlation coefficient r has ranged from 0.645 to 0.77, $P<.001$. Several studies, some overlapping, have also addressed the positive predictive value of specific TRV values for right heart catheterization–documented pulmonary hypertension, and these are summarized. Based on these studies, more than half of SCD patients with TRV ≥2.9 m/sec have pulmonary hypertension on right heart catheterization, whereas <15% of those with TRV 2.5 to 2.8 m/sec have pulmonary hypertension. However, performing right heart catheterizations only in SCD patients with TRV ≥2.9 m/sec leads to a failure to diagnose pulmonary hypertension in about 4 of 10 patients with the condition.

The accuracy of noninvasive diagnosis of pulmonary hypertension in SCD could be improved by a multimodality approach: the combination of TRV 2.5 to 2.8 m/s, NT-proBNP>164.5 pg/mL, and 6-minute walk distance <333 m had a positive predictive value of 62% with a false-negative rate of 7%. Therefore, SCD patients with TRV of 2.5 to 2.8 m/sec should be considered for right heart catheterization if there are other findings that suggest pulmonary hypertension, such as dyspnoea on exertion, increased brain natriureticpeptide, right ventricular hypertrophy seen byechocardiography, and/or limited exercise capability. Slightly more than half of the reported cases of right heart catheterization–documented pulmonary hypertension in SCD have precapillary pulmonary hypertension. The diagnosis of pulmonary arterial hypertension in patients without SCD traditionally includes the additional criterion of elevated pulmonary vascular resistance as defined by >3 Wood units. However, it is recognized that the elevation in cardiac output and reduced blood viscosity associated with SCD results in a lower baseline pulmonary vascular resistance than among nonanemic subjects and that SCD subjects with precapillary pulmonary hypertension often do not have elevation in pulmonary vascular resistance by the conventional definition. The American Thoracic Society's Ad Hoc Committee on Pulmonary Hypertension of Sickle Cell Disease recommended a revised definition of elevated pulmonary vascular resistance in SCD of >2 Wood units. It is hemodynamically possible to have both pre-and postcapillary pulmonary hypertension in the same patient, and patients with postcapillary pulmonary hypertension should bereevaluated after effective therapy for left ventricular dysfunction to determine whether elevation of the mean pulmonary artery pressure persists. Mortality with precapillary pulmonary hypertension is at least as high as mortality with postcapillary pulmonary hypertension. An analysis of the National Institutes of Health cohort indicated that mortality in SCD adults with pulmonary hypertension is driven primarily by the severity of their precapillary pulmonary vascular pathology: those who died had significantly higher mean pulmonary artery pressure, transpulmonary pressure gradient, and pulmonary vascular resistance13-15

2. Material and Methods

The study comprised of all diagnosed cases of haemoglobinopathies admitted in Medicine Department tertiary care centre between December 2017 to October 2019.

Research design
The researcher has adopted prospective cross sectional design to find correlation of pulmonary arterial hypertension (PAH) with patient of haemoglobinopathies.

Population
For the present study the population was all haemoglobinopathy patients admitted as well as outpatient in medicine department of tertiary care centre.

Sampling technique

For the present study non-probability purposive sampling method was used to select all diagnosed patients of hemoglobinopathy in medicine department tertiary care centre and who met the designed set of criteria.

Calculation of sample size
Calculated by formula: $Z^2 \times P \times Q/e^2$
where Z=constant whose value is 1.96 at 95% confidence interval
P= prevalence which is taken as 3%[16]
Q = 1-P
e= allowable error taken as 5%
The total sample size = 45
The sample size selected for this study was 50.

Criteria for sample selection
The following criteria were set for the selection of sample.

Inclusion Criteria:
1) Patients above the age of 12 years.
2) Patient diagnosed to have haemoglobinopathies.
3) Patient who gave consent for study.

Exclusion Criteria:
1) Patients with ischemic heart disease
2) Any structural heart disease in addition to haemoglobinopathies like congenital heart disease, rheumatic heart disease, tuberculous affections of heart.
3) History of any pulmonary diseases other than haemoglobinopathies complications.
4) Patients who declined inclusion in study or denied consent.

Baseline screening process and recording:
- Detail clinical history of patient.
- Thorough clinical examination
- Routine blood investigation-blood group, complete blood count, bleeding time, clotting time, blood sugar level
- Sickling Test.
- High performance liquid chromatography.
- 2D Echocardiography.

Procedure of data collection:
- After applying inclusion and exclusion criteria total cases were studied.
- Data was collected in the proforma on admission till discharge.
- Detailed case perform used which is already validated by department faculties for entering all details of patient.
- After written valid informed consent of patient, case proforma filled with details like name, age, sex, clinical features on arrival, previous significant histories contributing to the present condition (DM / HTN / H/o blood disorders), habits are recorded.
- After initial diagnosis, details regarding the status of the patient on admission with respect to vitals, sickling test and 2 D Echo were noted.

Study parameter evaluation: Cardiac status evaluations by 2D ECHO

1) Systolic Pulmonary Artery Hypertension was defined as PASP greater than 25 mm of Hg. Pulmonary artery systolic pressure (PASP) was calculated from the tricuspid regurgitation velocity
2) (TRV) and the estimated right atrial pressure (RAP) using the; Modified Bernoulli equation: PASP = 4 (TRV x TRV) + RAP[17]
3) The mean RAP was calculated according to the degree of collapse of the inferior vena cava with inspiration: 5 mmHg for a collapse of at least 50% and 15 mmHg for a collapse of less than 50%.
4) The reference ranges for mean pulmonary artery pressure are as follows
   - Normal –mPAP< 25 mm of Hg.
   - Mild PAH – mPAP 25-40 mm of Hg.
   - Moderate PAH – mPAP 41-55 mm of Hg.
   - Severe PAH – mPAP>55 mm of Hg

Data analysis

Analysis of data is done using SPSS version 20. Qualitative variables were expressed as frequency and percentage. Chi-square test, correlation coefficient, Logistic Regression, ANNOVA and MANCOVA was used to compare qualitative variables. Level of significance "P" value was evaluated, where P value < 0.05 was considered statistically significant.

3. Results

A study was undertaken to know pulmonary hypertension by 2D echocardiography in haemoglobinopathies in 50 cases, from December 2017 to October 2019 study period at tertiary care centre data were analysed using computer based SPSS software by frequency, percentage etc. . The p-value < 0.05 was considered as statistically significant

**Table 1:** Age & sex distribution of study population, n = 200

Age Group (in years)	Number	Percentage	Mean ± SD
12-20	15	30	
21-30	26	52	
31-40	4	8	25 ± 10.35
41-50	4	8	
51-60	1	2	
TOTAL	50	100	

Fig. 1: Age wise distribution of samples
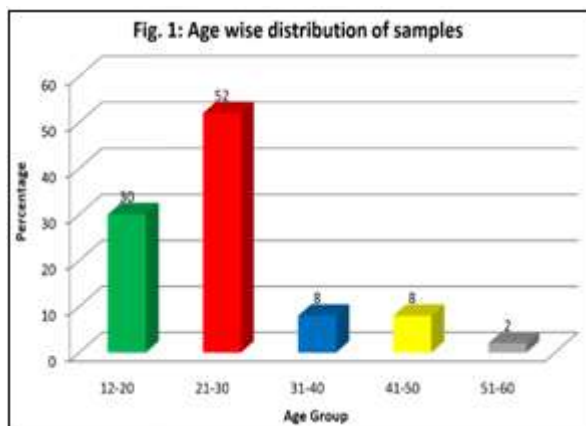
Maximum patients were in age group 21 to 30 (52%) Mean age score was 25 and standard deviation was 10.35.

Table 2: Sex wise distribution of study population

Sex	Number	Percentage
Male	18	36
Female	32	64

Fig.2: sex distribution of samples
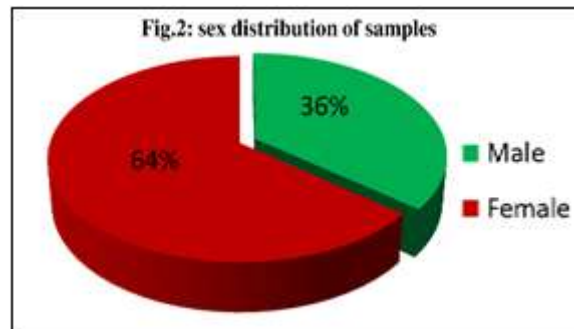
In my study female preponderance was observed i. e.64% (32 out of 50) were females. Mean age score for male was 24.89 and standard deviation was 10.65 whereas for female mean age score was 25.06 and standard deviation was 10.36.

Table 3: Haemoglobinopathies observed (sex wise), n = 50

Hemoglobinopathy	No. of cases		
	Male	Female	Total
Sickle cell anemia	07 (14 %)	13 (26 %)	20 (40 %)
Sickle cell trait	03 (06 %)	03 (06 %)	06 (12 %)
Beta thalassemia major	03 (06 %)	03 (06 %)	06 (12 %)
Beta thalassemia intermedia	01 (02 %)	03 (06 %)	04 (08 %)
Beta thalassemia trait	01 (02 %)	06 (12 %)	07 (14 %)
Sickle cell anemia + Beta thalassemia major	02 (04 %)	04 (08 %)	06 (12 %)
Beta thalassemia major + HbE Homozygous	01 (02 %)	00 (00 %)	01 (02 %)

Out of 50 patient 20 (40%) were having sickle cell anaemia while 06 (12%) were having Sickle cell trait, Beta thalassemia major &Sickle cell anemia + Beta thalassemia major respectively. Beta thalassemia trait was seen in 07 (14 %) of cases whereas Beta thalassemia intermedia was seen in 04 (08 %). Furthermore there was no significant association found between gender and sickle cell disease (p>0.05). maximum observed patients had sickle cell anemia. Only one patient had HBE + beta thalassemia.
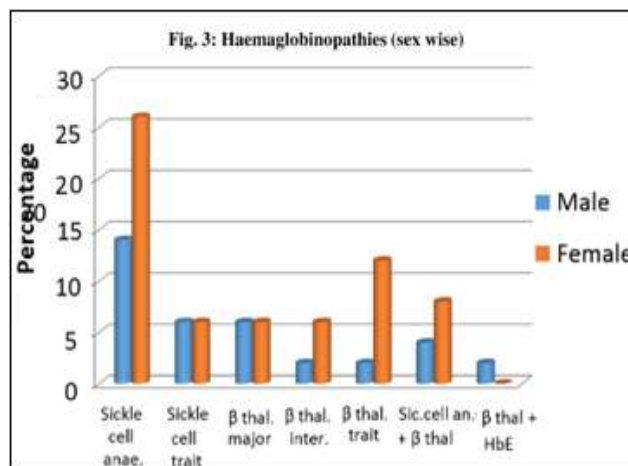

Fig. 3: Haemaglobinopathies (sex wise)

Table 4: HB levels observed in hemoglobinopathies

Hemoglobinopathy	Total cases	Hemoglobin (mg/dL)			Chi square	p value
		< 7	7.1-10	> 10.1		
Sickle cell disease	20	09 (45%)	09 (45%)	02 (10%)	3.489	0.240
Sickle cell trait	06	01 (16.66%)	00	05 (83.33%)	13.489	0.120
Beta thalassemia major	06	05 (83.33%)	01 (16.66%)	00	3.643	0.303
Beta thalassemia intermedia	04	04 (100%)	00	00	4.715	0.194
Beta thalassemia trait	07	02 (28.56%)	02 (28.56%)	03 (42.84%)	2.796	0.112
Sickle cell anemia + Beta thalassemia major	06	02 (33.33%)	04 (66.66%)	00	4.231	0.238
Beta thalassemia major + HbE Homozygous	01	01 (100%)	00	00	1.114	0.776
Total	50	24 (48%)	16 (32%)	10 (20%)		

Fig.4 : Hemoglobin level

■ < 7 mg/dL
■ 7.1-10 mg/dL
■ 10.1-12 mg/dL
■ Normal
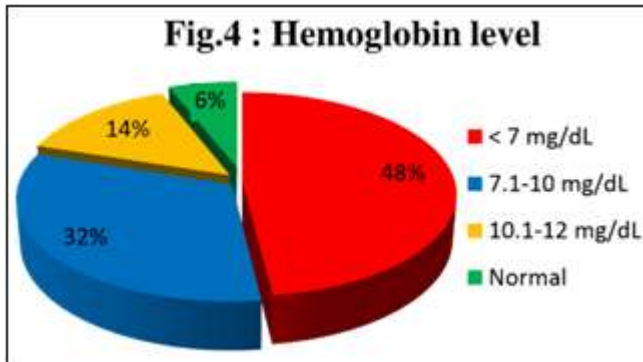
Out of 50 patients of haemoglobinopathies only 10 (20%) had Hb >10.1 gms % while 24 (48%) had severe anaemia Hb<7 gms %. Of those severe anaemia with sickle cell disease were 9 out of 20 (45%) and thalassemia major 4 out of 6 (81%) while sickle cell trait 33% had severe anaemia and beta thallasemia trait 28.4% had severe anemia.

Out of 50 patient 26 (52%) were having mild degree of PH, 4 % patient having moderate degree of PH whereas only 2 % patient having severe PH. Mean systolic pulmonary artery pressure (SPAP) was 25.80 and standard deviation was 9.06 Maximum patients of mild PH were having sickle cell anemia.

Furthermore there was no significant association found between systolic pulmonary artery pressure and sickle cell disease (p>0.05) except for Beta thalassemia major where p=0.011.

Table 5: Haemoglobinopathies in correlation to mean pulmonary artery

Hemoglobinopathy	Mean Pulmonary Artery Pressure mm Hg				Total	p value
	Normal (< 25 mmHg)	Mild (25-40 mmHg)	Moderate (4155mmHg)	Severe (>55mmHg)		
Sickle cell disease	6 (30%)	14 (70 %)	0	0	20	0.156
Sickle cell trait	3 (50%)	3 (50%)	0	0	6	0.915
Beta thalassemia major	1 (16.67%)	3 (50%)	1 (16.67%)	1 (16.67%)	6	0.011*
Beta thalassemia intermedia	2 (50%)	1 (25%)	1 (25%)	0	4	0.135
Beta thalassemia trait	6 (85.71%)	1 (14.29%)	0	0	7	0.093
Sickle cell anemia + Beta thalassemia major	3 (50%)	3 (50%)	0	0	6	0.25
Beta thalassemia major + HbE Homozygous	0	1 (100%)	0	0	1	0.815
Total	21 (42%)	26 (52%)	2 (4%)	1 (2%)	50	

Out of 50 patient 26 (52%) were having mild degree of PH, 4 % patient having moderate degree of PH whereas only 2 % patient having severe PH. Mean systolic pulmonary artery pressure (SPAP) was 25.80 and standard deviation was 9.06 Maximum patients of mild PH were having sickle cell anemia.

Furthermore there was no significant association found between systolic pulmonary artery pressure and sickle cell disease (p>0.05) except for Beta thalassemia major where p=0.011.

Table 6: Analysis of pulmonary hypertension with respect to agegroups n = 50

Age group	Mean Pulmonary Artery Pressure				Total	p value
	Normal (mPAP<25)	Mild (mPAP 25-40)	Moderate (mPAP 41-55)	Severe (mPAP> 55)		
10-20	5 (10%)	10 (20 %)	0	0	15	
21-30	10 (20%)	13 (26%)	02 (4%)	01 (2%)	26	
31-40	03 (6%)	01 (2%)	0	0	4	0.687
41-50	03 (6%)	01 (2%)	0	0	4	
51-60	0	1 (2%)	0	0	1	
Total	21 (42%)	26 (52%)	2 (4%)	1 (2%)	50	

There were only three patient (6%) of pulmonary hypertension. It also shows that the incidence of PH increased with the age of the patients in the study; 52% of patient the age group of 21-30 years had PH (4%). Also the correlation is not found significant (p > 0.05).

Table 7: Analysis of pulmonary hypertension with respect to Gender, n = 50

Gender	Mean pulmonary artery pressure in mm Hg				Total	p value
	Normal (mPAP<25)	Mild ((mPAP25-40)	Moderate (mPAP 41-55)	Severe (mPAP> 55)		
Male	7 (14%)	9 (18 %)	01 (2%)	01 (2%)	18	
Female	14 (28%)	17 (34%)	01 (2%)	0	32	0.565
TOTAL	21 (42%)	26 (52%)	2 (4%)	1 (2%)	50	

All three patients (6%) of pulmonary hypertension were males. It also shows that the no. of females (64%) was more than males (36%) in the study; also the correlation of gender to PH was found significant (p < 0.05).

Table no.18: Other echocardiographic findings in haemoglobinopathies

S. N.	Echocardiographic Findings	Male	Female	Total
1	Diastolic dysfunction	1	2	3
2	Systolic Dysfunction	0	0	0
3	Concentric LVH	0	1	1
4	Dilated left ventricle	0	0	0

in this study 4 patients having cardiac findings other than pulmonary hypertension. Out of 4 patients 3 patients were having diastolic dysfunction and 1 patient having Concentric LVH.

4. Discussion

In the present study table 1 shows maximum number of patients were in age group 21 to 30 (52%) which is relate to the findings in Jaya Pataki et al16 (2013) study in which majority of patients were in age group 14 to 20 years (46%). In contrary to this Atonally Melon et al (2015) 18 study conducted in USA was uncorrelated to age.

In my study table 2 shows female (64%) constitute more population than male (36%) by making female-male ratio 1.77: 1 which is relate to findings of the French registry which found a female-male ratio 1.9: 119. In contrary to this Jaya Pataki et al (2013) study male (68%) constitute more population than female (32%).

Sickle cell anaemia was found among 40% of samples in present study. There was no significant association was found between gender and hemoglobinopathies in the present study. However in Jaya Pataki et al (2013) 16 study 72% population was having sickle cell disease and 28 % was having sickle cell trait. Maharashtra, the sickle cell gene is widespread in all the eastern districts, also known as the Vidarbha region, in the Saputara ranges in the north and in some parts of Marathwada. The prevalence of sickle cell carriers in different tribes varies from 0 to 35 %20.

In the present study pulmonary hypertension with respect to age groups shows that most of the cases (52%) belongs to 21-30 years of age which correlates to findings of studies done by Aessopos et al (2015), Taher et al (2002), Derchi et al (2014). Hagar et al (2006), Morris et al (2011) where prevalence was mostly between 21 to 30 years of age.21-25

In the present study there were only 3 cases (6%) of moderate and severe PAH was found. In that 2 cases were suffering from beta thalassemia major (4%) and it was significantly associated with PAH (p<0.05) however in the study done by Fraidenburg DR et al (2016) 26 states that pulmonary hypertension in beta thalassemia major correlates with severity of hemolysis, yet in patients whose disease is well treated with chronic transfusion therapy, the development of pulmonary hypertension can be related to cardiac dysfunction and the subsequent toxic effects of iron overload rather than hemolysis.

In this study it is observed that total 4 patients were having cardiac manifestations other than pulmonary hypertension which was statistically not significant.

This cross-sectional, study was conducted with only 50 patients hence it could not derive proper boundaries for cardiac parameters. Larger sample strength would be needed to accurately decide the cut off points.

5. Conclusion

In the study period of 2 years only 50 patients presented to medicine department of this tertiary care centre. Of the total hemoglobinopathies maximum were sickle cell disease (40%) followed by beta thalassemia trait (14%). Maximum patients were in age group 21 to 30 years (52%). In my study female patients were more (64%) than male patients (16%). Haemoglobin <7gms% were observed in 24 (48%) patients while Hb 7-10 gms% in 16 (32%) patients. Out of 6 beta thalassemia major patients 5 patients had pulmonary hypertension, 1 had severe, 1 had moderate and 3 had mild pulmonary hypertension.

In sickle cell disease 14 patients out of 20 (70%) had mild pulmonary hypertension. No statistically significant difference was observed between males and females regarding development of pulmonary hypertension. This study highlights importance of screening for pulmonary hypertension in hemoglobinopathies to detect development of pulmonary hypertension at the earliest.

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