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EVALUATE AND COMPARE ABDOMINAL SONOGRAPHIC FINDINGS IN INDIVIDUALS WITH THALASSEMIA AND HEALTHY INDIVIDUALS

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Abstract

Background: *This study evaluates organs involvement pattern, liver size or echotexture, splenomegaly, ascites, gall bladder abnormalities, cortical echogenicity and renal alterations in thalassemia patients.*

Objective: *To evaluate and compare abdominal sonographic findings in individuals with thalassemia and healthy individuals.*

Methodology: *This study employs a cross-sectional comparative design, conducted at the Ruqayya Medical Centre and Fatimid Foundation, Lahore, utilizing a Toshiba Nemio XG ultrasound machine, to assess the prevalence and characteristics of abdominal organs among thalassemia and healthy individuals. A total sample size of 150 participants. The sampling technique adopted was convenient sampling, ensuring the inclusion of eligible participants during the study period.*

Results: *The study considered various demographic, clinical, and different abdominal organs-related variables to assess the prevalence and severity of thalassemia. Gender distribution included 81 females (54.0%) and 69 males (46.0%), with age groups divided into <12 years (33.3%), 13-18 years (58.7%), and above 30 years (8.0%). Liver size included homogeneous (12.0%), Heterogenous (82.0%), and Coarse (6.0%), Splenomegaly was reported by (67.6%), Gall bladder wall thickness status showed moderate (3.3%), severe (48.0%) and (28.0%) exhibited Sludge and (8.7%) had Calculi, Ascites in (31.3%). while (30.0%) experienced Mildly increased cortical echogenicity. In right Kidney group (36.0%) reported small, (33.3%) exhibited large right kidney, while left kidney group (42.0%) reported small left kidney, (32.0%) exhibited large left kidney.*

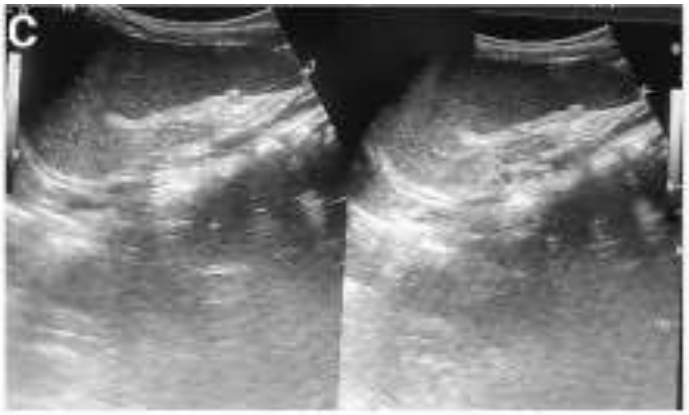
Conclusions *This study emphasizes the importance of routine abdominal ultrasound in early detection and management of organ complication in thalassemia. Regular monitoring may help to control diseases progression and improve patient's outcomes.*

INTRODUCTION

Thalassemia is a group of genetic conditions in which there is insufficient production of at least one of the globin chains, resulting in an imbalance in the production of globin chains. Damaged hemoglobin eventually results in anemia (1). The corresponding proteins form the adult hemoglobin molecule (HbA) which is a heterotetramer of two α and two β globin chains. α-Thalassemia resulting from mutations in the α-globin genes, it ranges from asymptomatic carrier states to severe hemoglobin H disease (2). β-Thalassemia Caused by mutations in the β-globin genes, this type includes β-thalassemia major (severe

form) and β-thalassemia minor (carrier state) (3). The severity of alpha and beta thalassemia is determined by the number of missing genes for either alpha (four) or beta globin (two). The Mediterranean, Africa, Pakistan, the Middle East, the Indian subcontinent, and Southeast Asia are all home to thalassemia. At least 300,000 babies worldwide are born with severe hemoglobinopathy each year. There were 1,310,407 cases of thalassemia worldwide in 2021. This trend highlights the importance of the early detection of comorbidities such as diabetes, hypertension, and cardiovascular diseases in thalassemia patients (6).

Thalassemia is a genetic blood disorder that can cause serious problems in the spleen, increased gut iron absorption, high liver iron accumulation, and kidney problems. Despite the widespread use of ultrasonography in the clinical evaluation of splenic enlargement, no one can agree on how to define splenomegaly in SCD patients (7). Some authors have used the spleen length to define splenomegaly, while others have used the spleen volume or index. Ultrasonography of the abdomen reveals that SCD patients' spleens vary in size (8)



In Figure 1.1, a longitudinal view of a six-year-old male presents with Hb thalassemia having an enlarged spleen (spleen length 9.9 cm).

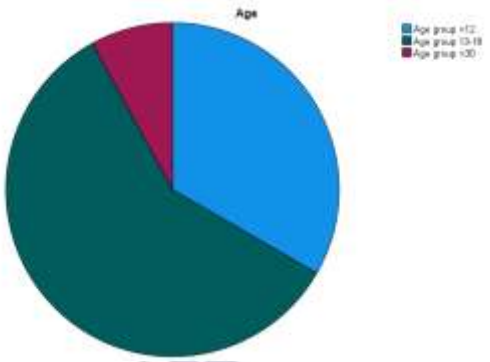
Problems with the production of hemoglobin, the oxygen-carrying protein in red blood cells, are caused by mutations in the genes that make the alpha- or beta-chains of hemoglobin (9). The pathophysiology of thalassemia involves a disruption in the balance of globin chain synthesis, leading to an imbalance in the alpha- and beta globin chains and subsequent abnormalities in red blood cell formation and function (10). Thalassemia is characterized by decreased globin chain synthesis, which causes varying degrees of anemia, early cell death, and ineffective erythropoiesis (11). Some of the undeveloped red blood cells enter the bloodstream. They are fragile and susceptible to hemolysis due to a defect in their membrane. In addition, their deformability has changed, and the spleen traps them and kills them with macrophages. The removal of platelets, white cells, and red cells as a result of this kind of spleen enlargement can lead to functional hypersplenism. Ineffective erythropoiesis, removal of abnormal cells by the spleen, and hemolysis all contribute to an anemia of variable severity (12).

The clinical onset of the first symptoms of severe and -thalassemia differs due to a physiologic switch in Hb levels during fetal and infant development (13). Due to the physiological inhibition of the g-globin genes that are responsible for the production of Hb F, the first clinical manifestation typically occurs between the ages of six months and two years. As a result of progressive splenomegaly and hepatomegaly or recurrent infections, these patients may present with severe anemia (Hb7g/dl), pallor, jaundice, irritability, inability to thrive, skeletal deformities, and abdominal enlargement. As a result, their survival depends on regular blood transfusions (14). All of the side effects, like too much iron, viruses, and damage to organs like heart failure, cirrhosis, and other endocrinopathies (15).

RESULTS:

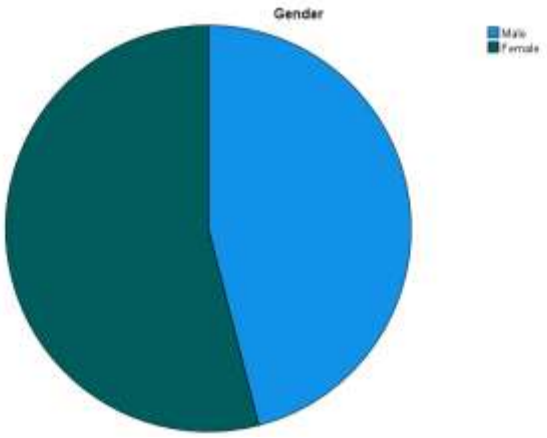
The data includes 150 participants categorized into three age groups. The <12 age group comprises 50 individuals (33.3%), while the 13-18 group is the largest, with 88 participants (58.7%). The remaining 12 participants (8.0%) fall into the age group above 30, making up 100% of the total sample.

Age		
	Frequency	Percent
Age group <12	50	33.3
Age group 13-18	88	58.7
Age group >30	12	8.0
Total	150	100.0



The data shows a total of 150 participants, with 69 males (46.0%) and 81 females (54.0%).

Gender		
	Frequency	Percent
Female	81	54.0
Male	69	46.0
Total	150	100.0



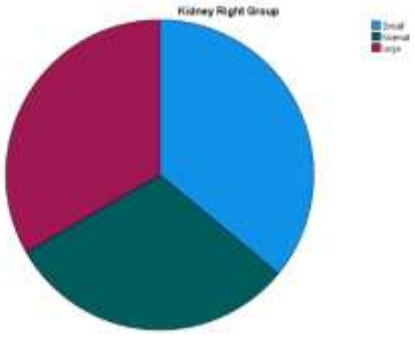
Out of 150 participants: 9 (6.0%) reported Coarse liver, 123 (82.0%) experienced Heterogenous liver, and 18 (12.0%) had Homogenous. While 39 (26.0%) reported Homogenous liver echotexture ,36 (24.0%) experienced Heterogenous while 75 (50.0%) had normal liver echotexture. 76 (50.7%) had normal

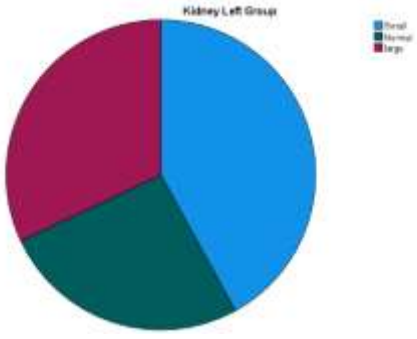
spleen size, while 11 (7.3%) exhibited moderate size, while 63(42.0%) experienced severe spleen size. 73 (48.7%) had normal GB wall thickness, 5 (3.3%) exhibited moderate size, while 72(48.0%) experienced severe Gall bladder wall thickness. 95 (63.3%) had no Sludge/Calculi, 42 (28.0%) exhibited Sludge and 13 (8.7%) had Calculi.

Liver size	Coarse liver	9	6.0	150
	Heterogeneous liver	23	82.0	150
	Homogeneous	18	12.0	150
Liver echotexture	Normal	75	50.0	150
	heterogeneous	36	24.0	150
	Homogeneous	39	26.0	150
Spleen size	Normal	76	50.0	150
	Moderate size	11	7.3	150
	Severe size	63	42.0	150
Gallbladder wall	Normal	73	48.7	150
	Moderate	5	3.3	150
	Severe	72	48.0	150
Sludge/ Calculi	Absent	95	63.3	150
	Sludge	42	28.0	150
	Calculi	13	8.7	150

Among 150 participants: In right kidney group, 46 participants (30.7%) had normal 54 participants (36.0%) reported small, while 50 participants (33.3%) exhibited large right kidney. In left kidney group, 39 participants (68.7%) had normal left kidney, 63 participants (42.0%) reported small left kidney, while 48 (32.0%) exhibited large left kidney.

Right Kidney Group	Small	54	36.0	150
	Normal	46	30.7	150
	Large	50	33.3	150
Left Kidney Group	Small	63	42.0	150
	Normal	39	26.0	150
	Large	48	32.0	150

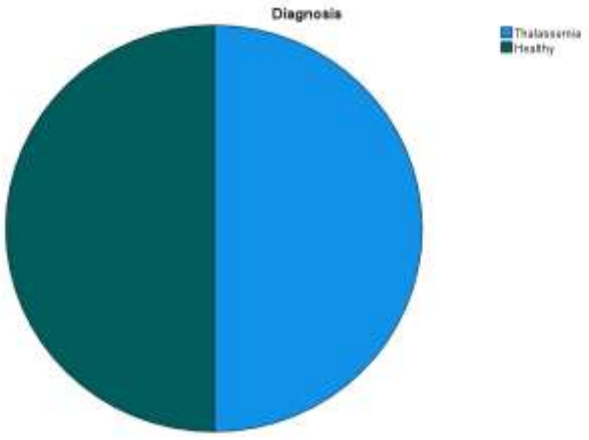




Out of 150 participants, 75 (50.0%) individuals had Thalassemia and 75 (50.0%) were Healthy.

Diagnosis		
	Frequency	Percent
Healthy	75	50.0
Thalassemia	75	50.0

Total	150	100.0



The frequency distribution of age shows that out of 150 participants, 50 (33.3%) belonged to the >12 age group, 88 (58.7%) were in the 13–18 age group, and 12 (8.0%) were above 30 years of age. 69 participants were male (46.0%) and 81 participants were female (54.0%), respectively.

In terms of liver echotexture, 36 participants (24.0%) exhibited heterogeneous liver patterns, 39 participants (26.0%) had homogeneous echotexture while 75 participants (50.0%) had coarse liver echotexture, according to a different classification. Regarding liver size 9 (6.0%) reported Coarse liver, 123 (82.0%) experienced Heterogenous liver, and 18 (12.0%) had Homogenous. In term of spleen size, 76 (50.7%) had normal spleen size, while 11 (7.3%) exhibited moderate size.

Regarding gall bladder wall thickness, 73 (48.7%) had normal, 5 (3.3%) exhibited moderate size, while 72(48.0%) experienced severe Gall bladder wall thickness. For sludge/calculi, 95 (63.3%) had no Sludge/Calculi, 42 (28.0%) exhibited Sludge and 13 (8.7%) had Calculi.

The frequence distribution of right kidney, 46 participants (30.7%) had normal 54 participants (36.0%) reported small, while 50 participants (33.3%) exhibited large right kidney. For left kidney, 39 participants (68.7%) had normal left kidney, 63 participants (42.0%) reported small left kidney, while 48 (32.0%) exhibited large left kidney. In addition, the sample was divided according to diagnosis, with 75 participants (50 percent) reported Thalassemia and 75 participants (50 percent) being healthy.

The distribution of kidneys pattern in the 13-18 age group exhibiting higher rates of thalassemia compared to the younger and older groups.This demonstrated that there was a distinct relationship between kidney size and Thalassemia status.

DISCUSSION.

The gender distribution in our study (male-to-female ratio 1:1.17) closely aligns with the findings of Al-Khabori M et al. (2023), who reported a higher proportion of females (male-to-female ratio 1:1.15) in thalassemia (15). This trend may reflect increased healthcare-seeking behavior among women. Conversely, Abbas et al. (2015) also observed a female-to-male ratio of approximately 1.2:1 in 150 thalassemia patients undergoing abdominal ultrasound (16).

Our findings are slightly higher for severe gall bladder wall thickness by 48.0% of participants, which is comparable to the 43% prevalence reported by McCarthy et al. (2024) among patients presenting with gallstones. while moderate levels are reported by 33.0% vs. 35% (17). This study's prevalence of gallbladder calculi, 8.7%, is higher than that of Ighodaro OE et al.'s 6% prevalence. (2022). This discrepancy may reflect population-specific differences, such as co-existing conditions (18). While the prevalence of sludge 28.0%, which is comparable to the 31.3% reported by Damayanti EL et al. (2024) (19). Splenomegaly, the proportion of participants experiencing severe spleen size 42% closely matches findings from Ladu AI et al. (2024),

who reported enlarge spleen size 41.3% (20). The distribution of moderate spleen size (23.9%) also reflects a balanced representation, aligning with Saif A et al. (2015), who reported similar proportions is 25% moderate (21). Severe spleen size was reported by 42.0% of participants in the above 30 age group, which is comparable to the 40% prevalence in above 32 age group reported by Eze CU et al. (2015) among patients presenting with sickle cell thalassemia, while moderate spleen size is different 7.3% vs. 15.0% (22).

Mild Ascites was reported by 31.3% of participants, which is comparable to the 40% prevalence reported by Fatigati C et al. (2024) among patients presenting with hepatomegaly Sickle cell thalassemia. Our findings are slightly lower for mild Ascites compared to their report (23). Mild ascites although not common, which may reflect early stages of portal hypertension or chronic liver dysfunction reported by Rodeghiero F et al. (2012) (24).

Liver size, the prevalence of coarse liver 6.0% is slightly lower than the 13% reported by Ferraioli G, et al. (2016) in a comparable cohort. This could reflect differences in participant characteristics (25). Our findings slightly different, Heterogenous 16% and coarse liver 47.0% predominating in the teen age. Mangia A, et al. (2020) reported liver size and echotexture showed marked age-related alterations, with heterogenous 34% and coarse textures 28% predominating in older participants. Transfusion-dependent thalassemia patients have reported hepatic changes consistent with these findings (26). The prevalence of mildly increased renal echogenicity is 30.0%, enlarge right kidney 33.3% and left kidney is 32.0% aligns closely with McCarville MB et al. (2011), who reported a prevalence of mild renal echogenicity 35.0%, enlarge right kidney 29.3% and left kidney is 36.0%. The slight variability in rates may stem from variations in population demographics (27). While Patel R et al. (2019) reported the prevalence of mildly increased echogenicity 23%, kidneys were small 10% and enlarge 11 of the thalassemia patients (28). This discrepancy may reflect population-specific differences, such as co-existing conditions. The clustering of enlarge left or right kidney rates among thalassemia participants in the 13-18 age group underscores the need for early intervention strategies targeting this demographic. Furthermore, the comparable prevalence rates observed across multiple studies reinforce the generalizability of these findings, emphasizing the relevance of renal ultrasonography, particularly for middle-aged thalassemic individuals.

Conclusion:

This study emphasizes the importance of routine abdominal ultrasound in early detection and management of organ complication in thalassemia. Regular monitoring may help to control diseases progression and improve patient's outcomes.

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