Original Article

Epidemiological Insights into Hemoglobinopathies in Basrah, Southern of Iraq

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ABSTRACT

Objective: (1) To elicit evidence of new case detection in Basrah regarding hemoglobinopathy. (2) To study certain characteristics of the newly diagnosed cases. (3) To study characteristics of the newly registered patients in context of the premarital screening program issued since 6 years (4) To detect if the evidence of hemoglobinopathy within the families acts as a deterrent for the families from having further effected children.

Study Design: Retrospective, single-center analysis

Place and Duration of Study: This study was conducted at the Basrah Center for Hereditary Blood Diseases, Basrah Iraq from 1st January 2024 to 31st December 2024.

Methods: This retrospective, single-center analysis was conducted at Basrah Center for Hereditary Blood Diseases, Basrah Iraq vide letter No.18 dated 5th April 2023 and 508 individuals diagnosed with hemoglobinopathies were enrolled.

Results: Sickle cell anemia and sickle β-thalassemia accounted for 31.3% and 34.8% of cases, respectively. Gender distribution was nearly equal across all types, reflecting the autosomal recessive inheritance pattern. Consanguineous marriage was reported in 62% of cases, with the highest familial relation seen among sickle cell anemia patients. Familial clustering was evident, with 144 patients having at least one affected sibling. Educational attainment was low, with 45.5% of patients being illiterate and only 5.1% holding a college degree.

Conclusion: The strong influence of genetic, socio-cultural, and educational factors on hemoglobinopathy prevalence. The high rates of familial clustering and consanguinity highlight the urgent need for enhanced public health interventions, including premarital screening, genetic counseling and community education.

Key Words: Hemoglobinopathies, Epidemiology, Premarital Screening

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INTRODUCTION

The most prevalent single gene illnesses in the world are known as hemoglobinopathies, or disorders of hemoglobin.[1,2] Two primary classifications exist for hemoglobinopathies: thalassemia syndromes, and sickle cell diseases which include 2 main variants, homozygous sickle cell anemia and heterozygous sickle cell disease which may include many variants like S/β°, S/β+, S/D.[3]

The World Health Organization estimates that hemoglobinopathies affect about 5% of the human population and that 300,000–400,000 infants are born each year with extremely dangerous hemoglobinopathies.[4,5]

In general, one of the last recent studies indicates that the prevalence of thalassemia, through the data within 16 thalassemia centers in Iraq, was 37.1 per 100,000 in 2015. Between 2010 and December 2015, 11,165 cases of thalassemia were diagnosed, accounting for 66.3% of all hereditary anemia cases that were registered at these facilities. Which consider a significant rate with the need of attention and awareness to this hereditary disease and its burdens on our community.[5]

On the governorates distribution, a lot of studies had been held to elicit the prevalence within their population although separately. For instance, a study in the thalassemia center of Al-Najaf governorate, and by using the collected data of 1,122 patients from October 2019 to March 2020, it concluded that beta thalassemia is the number one diagnosed condition among the hereditary hematologic disorders and accounting for 33.15% of the patients registered in the center in duration of the study. In comparison, SCD account only for 9%.[6]

Another study in Erbil, northern Iraq, also found that beta thalassemia is the dominant condition among the hemoglobinopathies within their population, with a percentage as high as 78.71%. While the SCD account only for 6.23%. Overall, the peak age was between 6-15

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years account for 44.45%, following by 23.20% for the age group from 1-5 years. The 6-year study showed a steadily increase with all the hemoglobinopathies cases each year.[7]

The sickle-cell disease and thalassemia major from Iraq in the early 1960s, these hereditary disorders gained national recognition as serious health issues, leading to the establishment of centers for their management across the nation.[8]

In Basra Governorate, Southern of Iraq, whilst that the carrier state for both thalassemia (4.6%) and the frequencies of sickle cell trait prevalence vary greatly in different regions of Iraq (0.06–0.07%) in the North whilst it is in its extreme maximization (6.5%) in Basrah.[9]

The time trend of the newly registered patients in Basrah did show an accelerated increment in 2019, with the registration of about 606 new patients differing slightly from that of thalassemia. Iraqi trend for the last five years was published in an Iraqi study in which a steady increase did exist.[10,11] Again the most predominant newly registered were male gender, sickle cell diseases, patients from peripheries and below five years age groups.

METHODS

This retrospective study that had been conducted depending on the patient's records and center data bases had been obtained from the BCHBD after a written approval, data had been processed as numbers and percentages for obtaining the study objectives in highlighting the current situation for hemoglobinopathy in Basrah governorate beside the new patient registry characteristics. All hemoglobinopathy cases registered from January 2024 till end of December 2024 in Basrah center for hereditary blood diseases, hereditary bleeding disorders, query hereditary natures and odd diagnosis had been excluded. The data was entered and analyzed through SPSS-25.

RESULTS

There were 51.38% females and 48.62% males. Sickle thalassemia is the most common condition, accounting for 34.84% of all cases (n=177), with nearly equal distribution between females (17.52%) and males (17.32%). Sickle cell anemia follows closely with 31.30% of cases (n=159), also showing a slight female predominance (16.93%) compared to males (14.37%) These findings suggest that sickle-related disorders are highly prevalent in the studied group and affect both sexes similarly (Table 1).

62.20% of new cases (316 out of 508) occurred in individuals with consanguineous (relative) parents, while 37.80% (192) were from non-consanguineous unions. Sickle cell anemia follows with 31% (n=159) also showing a high number of cases among relatives (107 vs. 52) which suggests a notable genetic component possibly amplified by consanguinity (Table 2).

This raise a fact that 40.75% of the new registered were from an effected families with a complete knowledge of the disease inheritance in their families underscoring the hereditary nature of hemoglobinopathies. Sickle thalassemia (34.84%). The highest number of familial cases: 57 patients (11.22%) had one affected sibling, and 24 patients (4.72%) had two. A small but notable number had three or four affected siblings, indicating strong familial clustering in some households. Similarly, 44 patients (8.66%) had one affected sibling, and smaller percentages had two or more affected siblings. Still, a significant portion (19.69%) had no other affected siblings, suggesting it can also occur in families without known history. 59.25% (n=301) of the registered patients had no affected siblings, suggesting either sporadic inheritance or that the patient is the first diagnosed in the family.28.35% (n=144) had one affected sibling, while a smaller proportion had two (8.66%), three (2.36%), or four (1.38%) affected siblings. This raise a fact that 40.75% of the new registered were from an effected families with a complete knowledge of the disease inheritance in their families underscoring the hereditary nature of hemoglobinopathies. Sickle thalassemia (34.84% of total cases). The highest number of familial cases: 57 patients (11.22%) had one affected sibling, and 24 patients (4.72%) had two. A small but notable number had three or four affected siblings, indicating strong familial clustering in some households. Sickle cell anemia (31.30%) similarly, 44 patients (8.66%) had one affected sibling, and smaller percentages had two or more affected siblings. Still, a significant portion (19.69%) had no other affected siblings, suggesting it can also occur in families without known history. Thalassemia's (α and β types) β-thalassemia major and intermedia mostly appear in families with no affected siblings (5.12% and 3.74%, respectively). However, 6–8 patients per type had one or more affected siblings, reflecting known autosomal recessive inheritance. Fanconi anemia (1.38%), despite being rare, more than half of the cases had affected siblings (4 out of 7), suggesting strong genetic clustering in families with this condition. Nearly 41% of patients had at least one affected sibling, confirming the significant genetic and familial aspect of hemoglobinopathies (Table 3).

Illiteracy is the most dominant category, accounting for 45.47% (n=231) of patients, followed by primary education at 36.61% (n=186).Only 4.13% (n=21) of patients are college graduates, and 0.98% (n=5) institute graduates (Table 4).

70.67% of patients (n=359) are from peripheral areas, while only 29.33% (n=149) are from urban centers. This indicates a notably higher burden of hemoglobinopathies in peripheral areas, possibly due to

a combination of genetic, socioeconomic, and healthcare access factors. The significant concentration of cases in peripheral regions suggests potential factors (Table 5)

The highest proportion of patients (32.28%) was under 1 year old, indicating early onset and possibly neonatal screening or early symptom presentation. Children aged 1-5 years (22.83%) and 6-15 years (13.78%) make up the next largest groups, suggesting that early childhood remains the most common age for detection These disorders also show a small proportion of late diagnoses, with 7-9 patients over age 50 possibly due to milder forms or limited access to diagnosis in earlier life. No cases registered in individuals over 16 years, which aligns with early manifestation of physical or hematological symptoms (Table 6).

Sickle-related disorders (sickle cell anemia and sickle thalassemia) were the most frequent, together comprising over 66% of cases. Blood groups O+ and A+ were the most common among patients across all diagnoses (Table 7).

Table No.1: Categories of hemoglobinopathies towards sex

Type of hemoglobinopathy	Male	%	Female	%	Total	%
β. thalassemia major	19	3.74	15	2.95	34	6.69
β. thalassemia intermedia	16	3.15	17	3.35	33	6.50
α thalassemiaα	32	6.30	33	6.50	65	12.80
Sickle cell anemia	73	14.37	86	16.93	159	31.30
Sickle thalassemia	88	17.32	89	17.52	177	34.84
Fanconi anemia	3	0.59	4	0.79	7	1.38
Others	16	3.15	17	3.35	33	6.50
Sum	247	48.62	261	51.38	508	100.00

Table No.2: Statistics on new registered patients according to kinship

Type of hemoglobinopathy	Relatives	%	Not relatives	%	Total	%
β. thalassemia major	28	7.0	6	7.0	34	7.0
β. thalassemia intermedia	22	6.0	11	6.0	33	6.0
α thalassemiaα	43	13.0	22	13.0	65	13.0
Sickle cell anemia	107	31.0	52	31.0	159	31.0
Sickle thalassemia	93	35.0	84	35.0	177	35.0
Fanconi anemia	6	1.0	1	1.0	7	1.0
Others	17	6.0	16	6.0	33	6.0
Sum	316	100.0	192	100.0	508	100.0

Table No.3: Newly registered patients according to the number of affected saplings in the family

Type of hemoglobinopathy	Effected spilling	%	Effected spilling	%	Effected saplings	%	Effected saplings	%	Effected saplings	%	Total	%
β. thalassemia major	6	1.18	2	0.39	-	-	-	-	26	5.12	34	6.69
β. thalassemia intermedia	8	1.57	4	0.79	1	0.20	1	0.20	19	3.74	33	6.50
α thalassemiaα	10	1.97	2	0.39	-	-	-	-	53	10.43	65	12.80
Sickle cell anemia	44	8.66	11	2.17	3	0.59	1	0.20	100	19.69	159	31.30
Sickle thalassemia	57	11.22	24	4.72	8	1.57	5	0.98	83	16.34	177	34.84
Fanconi anemia	4	0.79	-	-	-	-	-	-	3	0.59	7	1.38
Others	15	2.95	1	0.20	-	-	-	-	17	3.35	33	6.50
Sum	144	28.35	44	8.66	12	2.36	7	1.38	301	59.25	508	100.0

Table No.4: Statistics on newly registered patients according to educational attainment

Type of hemo-globinopathy	Illiterate	%	Primary	%	Secondary	%	Preliminary	%	Institute raduate	%	College aduate	%	Total	%
β. thalassemia major	20	3.94	11	2.17	2	0.39	1	0.20	-	-	-	-	34	6.69
β. thalassemia intermedia	25	4.92	8	1.57	-	-	-	-	-	-	-	-	33	6.50
α thalassemiaα	21	4.13	30	5.91	11	2.17	2	0.39	-	-	1	0.20	65	12.80
Sickle cell anemia	68	13.39	53	10.43	20	3.94	6	1.18	2	0.39	10	1.97	159	31.30

Sickle thalassemia	74	14.57	69	13.58	14	2.76	8	1.57	3	0.59	9	1.77	177	34.84
Fanconi anemia	5	0.98	2	0.39	-	-	-	-	-	-	-	-	7	1.38
Others	18	3.54	13	2.56	1	0.20	-	-	-	-	1	0.20	33	6.50
Sum	231	45.47	186	36.61	48	9.45	17	3.35	5	0.98	21	4.13	508	100.0

Table No. 5: Statistics on newly registered patients by residential area

Type of hemoglobinopathy	Centre	%	Periphery	%	Total	%
β. thalassemia major	8	1.57	26	5.12	34	6.69
β. thalassemia intermedia	12	2.36	21	4.13	33	6.50
α thalassemiaα	24	4.72	41	8.07	65	12.80
Sickle cell anemia	41	8.07	118	23.23	159	31.30
Sickle thalassemia	55	10.83	122	24.02	177	34.84
Fanconi anemia	1	0.20	6	1.18	7	1.38
Others	8	1.57	25	4.92	33	6.50
Sum	149	29.33	359	70.67	508	100.0

Table No. 6: Distribution of various hemoglobinopathies based on age group

Type	<1 year	%	5 years	%	15 years	%	25 years	%	50 years	%	>50 years	%	Total	%
β. Thalassemia major	7	1.38	15	2.95	2	0.39	3	0.59	5	0.98	2	0.39	34	6.69
β.thalassemia intermedia	22	4.33	8	1.57	-	-		-	1	0.20	2	0.39	33	6.50
α thalassemiaα	1	0.20	17	3.35	18	3.54	11	2.17	16	3.15	2	0.39	65	12.80
Sickle cell anemia	-	-	58	11.42	40	7.87	20	3.94	34	6.69	7	1.38	159	31.30
Sickle thalassemia	4	0.79	56	11.02	41	8.07	29	5.71	38	7.48	9	1.77	177	34.84
Fanconi anemia	-	-	2	0.39	5	0.98	-	-	-	-	-	-	7	1.38
others	-	-	8	1.57	10	1.97	7	1.38	5	0.98	3	0.59	33	6.50
Total	34	6.69	164	32.28	116	22.83	70	13.78	99	19.49	25	4.92	508	100.00

Table No.7: Blood group distribution among new hemoglobinopathy cases

Diagnosis	O^-	O^+	AB^-	AB^+	B^-	B^+	A^-	A^+	Total
β-thalassemia major	2 (0.4%)	14 (2.8%)	1 (0.2%)	6 (1.2%)	1 (0.2%)	9 (1.8%)	1 (0.2%)	3 (0.6%)	37 (7.3%)
β-thalassemia intermedia	1 (0.2%)	14 (2.8%)	1 (0.2%)	1 (0.2%)	1 (0.2%)	4 (0.8%)	1 (0.2%)	11 (2.2%)	34 (6.7%)
α-thalassemia	2 (0.4%)	20 (3.9%)	1 (0.2%)	3 (0.6%)	1 (0.2%)	18 (3.5%)	2 (0.4%)	19 (3.7%)	66 (13.0%)
Sickle cell anemia	9 (1.8%)	54 (10.6%)	2 (0.4%)	10 (2.0%)	6 (1.2%)	33 (6.5%)	3 (0.6%)	42 (8.3%)	159 (31.3%)
Sickle thalassemia	12 (2.4%)	71 (14.0%)	1 (0.2%)	10 (2.0%)	1 (0.2%)	41 (8.1%)	1 (0.2%)	41 (8.1%)	178 (35.0%)
Fanconi anemia	2 (0.4%)	1 (0.2%)	-	1 (0.2%)	-	1 (0.2%)	-	2 (0.4%)	7 (1.4%)
Others	1 (0.2%)	9 (1.8%)	1 (0.2%)	4 (0.8%)	1 (0.2%)	7 (1.4%)	1 (0.2%)	11 (2.2%)	35 (6.9%)
Total	29 (5.7%)	175 (34.4%)	7 (1.4%)	35 (6.9%)	11 (2.2%)	113 (22.2%)	9 (1.8%)	129 (25.4%)	508 (100.0%)

DISCUSSION

Sickle cell anemia and sickle β-thalassemia accounted for the largest proportion of cases (31.3% and 34.8%, respectively). This high prevalence is in line with studies from the Middle East and North Africa (MENA) region, where sickle cell disorders are endemic due to historical malaria exposure and high consanguinity rates as in WHO publications, Bahrain[12] and Saudi Arabia.[13] Gender distribution across types was almost balanced, reflecting the autosomal recessive nature of these conditions. This is consistent with studies from Iran and India that observed similar gender ratios like what found in studies done in Iran, Pakistan and West Bengal.[14]

Kinship analysis showed that 62% of new cases came from consanguineous marriages, with the highest familial relation observed among sickle cell anemia (107 out of 159 cases). This supports extensive literature showing that consanguinity remains a major risk factor for autosomal recessive disorders like hemoglobinopathies in regions with traditional intra-familial marriage patterns a similar results found by Tadmouri et al[15] and Denic et al.[16]

This study showed that 144 patients had at least one affected sibling, indicating strong familial clustering. This pattern is frequently documented in hemoglobinopathy registries and reinforces the need for family-based screening and genetic counseling programs a thing elicited also by Cousens et al[17] and Grosse et al.[18]

There is an inverse relationship between educational level and disease prevalence: 231 patients (45.5%) were illiterate, while only 26 (5.1%) had college or institute degrees. Low health literacy has been previously associated with poor awareness about premarital screening and genetic counseling services in Saudi Arabia and Iran.[19] This underscores the importance of targeted awareness campaigns, especially in rural or underserved areas.

Approximately 29.3% of the registered patients were from rural areas. These findings are similar to those from regional surveys where rural residents have higher disease burden due to limited access to diagnostic services and genetic counseling a corresponding results found by Weatherall.[20]

The clustering of cases in consanguineous families with low education levels emphasizes the urgent need to integrate nationwide prevention programs. Premarital screening, early genetic counseling, and newborn screening have been successful in reducing incidence in countries like Cyprus, Iran and Saudi Arabia.[21]

Contrasting findings were reported in a study from India, where B+ was the most common blood group among β-thalassemia patients, followed by O+ and A+, a distribution potentially influenced by regional genetic variation and population structure.[22]

CONCLUSION

Hemoglobinopathies in the region are shaped by genetic, cultural, and socio-economic factors. A significant proportion of new registrants came from families with previously registered cases, reflecting a pattern of cognitive impairment among patients' families about the possibility of recurrence. Continued implementation of education-based and community-driven screening programs is crucial to reduce the incidence and burden of these diseases.

Author's Contribution:

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Final Approval of version:	All the above authors
Agreement to accountable for all aspects of work:	All the above authors

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