Volume 3, Number 3, 2026
Pages 719-727
Bioscience Insights
ISSN: 3080-3489 (Print)
ISSN: 3080-3497 (Online)
BiosciInsights.com

BETA-THALASSEMIA CARRIER FREQUENCY AND HAEMATOLOGICAL PROFILES AMONG INDIVIDUALS SCREENED IN NAROWAL, PUNJAB, PAKISTAN: A RETROSPECTIVE STUDY

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https://doi.org/10.5281/zenodo.23532091

Article Information

Article History

Received:	June 19, 2026
Revised:	August 19, 2026
Accepted:	September 07, 2026
Available Online:	October 21, 2026

Keywords:
Thalassemia, Carrier frequency, Haematological profiles, Narowal

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Abstract

Beta-thalassemia is a major inherited blood disorder that poses a significant global health burden, particularly in regions with high rates of consanguineous marriage such as Pakistan, where information on carrier frequency in rural communities remains limited. A retrospective analysis was conducted using 565 laboratory records from the thalassemia prevention programme at DHQ Hospital, Narowal, Punjab, Pakistan, covering January 2020 to November 2024. The study aimed to describe the frequency of β-thalassemia carriers and their haematological characteristics among individuals screened. Among these screened individuals, 122 (21.59%) were classified as carriers and 2 (0.35%) as having thalassemia major; these figures should not be interpreted as district-wide population prevalence because the sample comprised screening attendees. Carriers had lower MCV (64.97 vs. 81.92 fL) and MCH (18.76 vs. 26.04 pg), and higher HbA2 (5.32% vs. 2.57%) than non-carriers. Among the 85 advice entries summarized, family screening and genetic counselling and serum ferritin testing each accounted for 27 (31.76%), while haematinic supplementation accounted for 13 (15.29%). The findings support strengthening accessible screening, standardized CBC and haemoglobin-analysis protocols, genetic counselling, and community awareness. Future studies should include representative samples from surrounding districts to assess regional variation and the effectiveness of interventions.

INTRODUCTION

Beta-thalassemia is a significant inherited blood disorder that represents a major global health burden (Aksu & Unal, 2021). It is characterized by reduced or absent synthesis of the beta-globin chains of

haemoglobin, which are essential for normal red blood cell function (Muncie & Campbell, 2009). The condition ranges from the usually asymptomatic carrier state, known as thalassemia trait or minor, to severe, transfusion-dependent thalassemia major (Cao & Galanello, 2010). Imbalanced globin-chain production causes ineffective erythropoiesis and haemolytic anaemia; severe disease and repeated transfusions can also lead to iron overload complications (Porter & Garbowski, 2014; Fibach & Rachmilewitz, 2017).

Beta-thalassemia is generally inherited in an autosomal recessive pattern: an affected child usually inherits a pathogenic HBB variant from each parent (Thein, 2018). Carriers typically have one affected copy of the gene and are usually healthy, although they can transmit the variant to their children (Unissa et al., 2018). When both parents carry a pathogenic β-thalassemia variant, each pregnancy has a 25% probability of an affected child, a 50% probability of a carrier child, and a 25% probability of a child who inherits neither familial variant (Galanello & Origa, 2010). This inheritance pattern supports offering carrier testing and genetic counselling before marriage or pregnancy.

The burden of beta-thalassemia in Pakistan is an important public-health concern. In a study of families affected by β-thalassemia in Punjab, 96% of the families were reported to be consanguineous (Khan et al., 2023). Such findings describe families affected by the condition and should not be interpreted as the prevalence of consanguinity in the general population. Consanguinity can increase the probability that both parents carry the same recessive variant, increasing the risk of an affected child (Aziz et al., 2024).

Awareness of thalassemia genetics and the value of premarital and prenatal screening remain important in the general population (Yousaf et al., 2021). Rural healthcare challenges, including limited infrastructure, access to diagnostic equipment, and access to specialized services, may further complicate screening and counselling (Majrooh et al., 2015). These barriers can widen the gap between prevention needs and the availability of screening and counselling services, potentially contributing to affected births (Ahmed et al., 2017; Durrani et al., 2023).

The aim of the present study was to describe the frequency of β-thalassemia carriers and their haematological profiles among individuals screened in Narowal District, Punjab, Pakistan. The findings may help inform local prevention planning, genetic counselling, and public awareness. Because the study used screening records rather than a representative population sample, its estimates should be interpreted as applying to the screened individuals.

MATERIALS AND METHODS

This retrospective study used diagnostic reports from the Prime Minister's Thalassemia Prevention Programme (PTPP) at the District Headquarter (DHQ) Hospital in Narowal, Pakistan. Reports from samples screened between January 2020 and November 2024 were reviewed.

Study population and area

The study was conducted in Narowal District, in northeastern Punjab, Pakistan (Figure 1). The district includes urban and rural populations. The dataset comprised 565 diagnostic reports from individuals who presented for thalassemia screening under the programme. A population-based sampling frame was not described; therefore, the screened group cannot be assumed to represent the district population.

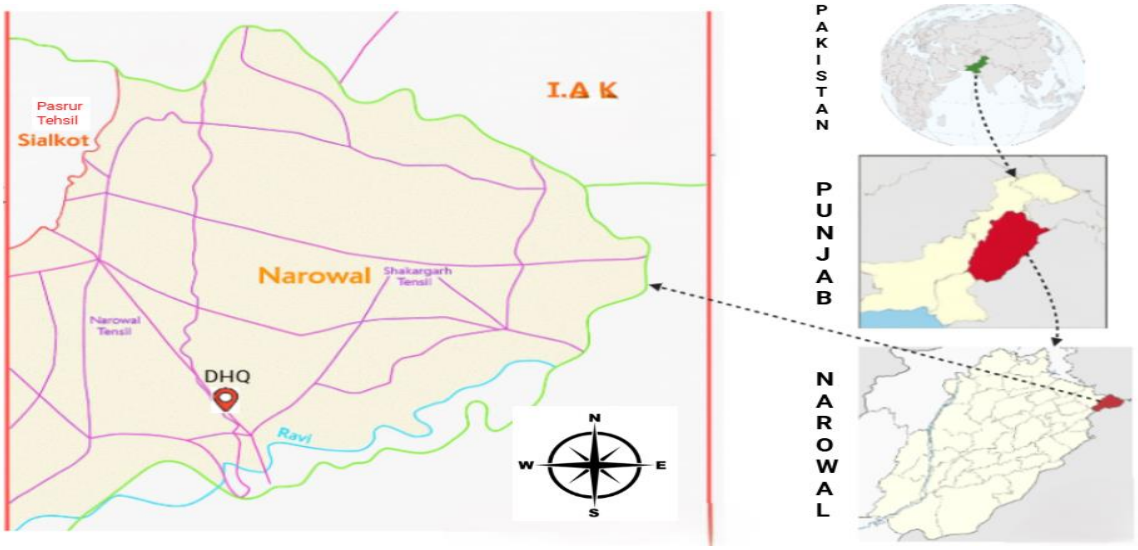


Figure 1: Map of District Narowal showing the DHQ (study area)

Table 1: Coordinates of the study area

Sr. No.	Area Name	Latitude	Longitude
1	District Headquarter Hospital (DHQ), Narowal	32.0992° N	74.8839° E

All lab tests were done under the standard PTPP protocol. To determine the most significant indices such as the level of haemoglobin, mean corpuscular volume (MCV) and mean corpuscular haemoglobin (MCH), a complete blood count (CBC) was carried out on an automated haematology analyser. Electrophoresis of haemoglobin was done on all the samples and the concentration of HbA and HbA2, as well as HbF, was determined.

An HbA2 level above 3.5%, often accompanied by reduced MCV (<80 fL) and MCH (<27 pg), was used to identify β-thalassemia carriers (trait). Individuals with HbA2 within the reference range and normal red-cell indices were categorized as non-carriers. Individuals with clinical and laboratory profiles consistent with thalassemia major were classified separately. Data from 565 diagnostic reports were manually extracted and compiled in Microsoft Excel. The data included CBC parameters, haemoglobin-analysis results, and clinical recommendations. Records were checked for completeness, and duplicates were removed before analysis. The frequencies of β-thalassemia trait and thalassemia major were calculated as percentages of the total screened sample. Descriptive statistics were used to summarize haematological parameters across the non-carrier, carrier, and thalassemia-major groups.

RESULTS

A total of 565 individuals were screened, of whom 122 were classified as β-thalassemia carriers (21.59%). Two individuals (0.35%) were classified as having thalassemia major (Table 2 and Figure 2).

Table 2: Frequency of β-thalassemia among screened individuals

Total Number of Individuals:	565
Number of Carriers	122
β-Thalassemia carrier frequency among screened individuals	21.59%
Number of Thalassemia Major	2
Thalassemia major frequency among screened individuals	0.35%

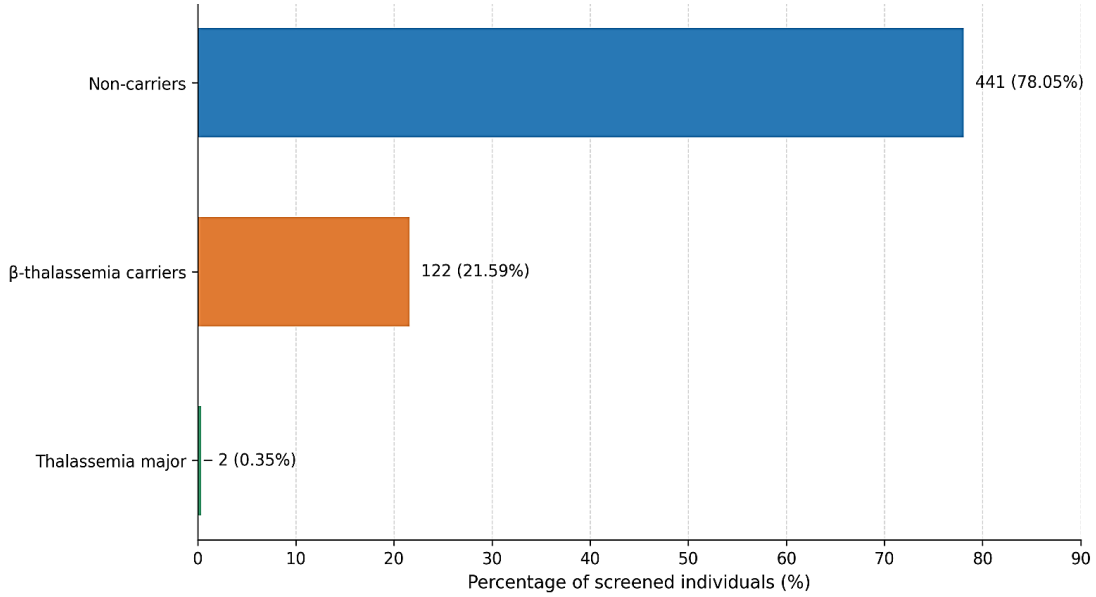


Figure 2: Frequency of β-thalassemia among screened individuals

Haematological indices differed descriptively between carriers and non-carriers: mean HGB was 10.87 g/dL in carriers versus 12.73 g/dL in non-carriers; MCV was 64.97 versus 81.92 fL, and MCH was 18.76 versus 26.04 pg. Haemoglobin analysis showed higher HbA2 (5.32% vs. 2.57%) and lower HbF (2.16% vs. 3.19%) in carriers. The two patients classified as having thalassemia major had severe anaemia (mean HGB: 4.65 g/dL) and elevated HbF (reported range: 51.9–53.1%). These findings are summarized in Table 3.

As shown in Table 3, platelet count was higher in carriers (327.10 × 10^3/μL) than in non-carriers (298.91 × 10^3/μL). RDW was also higher in carriers (17.23%) than in non-carriers (15.06%).

Table 3: Comparative haematological profile

CBC and haemoglobin-analysis parameter	β-Thalassemia non-carriers (comparison group)	β-Thalassemia carriers (trait)	Difference in Carriers (Carriers-Normal)	Thalassemia Major (affected patients)	Difference in Majors (Majors-Normal)
White blood cells (WBCs) (10^3/μL)	7.5-8.3	7.7-8.7	0.42	10.5-12	3.38
Red blood cells (RBCs) (10^6/μL)	4.2-4.9	4.9-5.9	0.95	1.5-2.2	-2.75
HGB (g/dL)	11.5-12.8	9.8-10.9	-1.86	4.1-4.7	-8.08
HCT (%)	38.8-39.8	36.5-37.7	-2.16	13.9-14.8	-25.07
MCH (pg)	25.1-26.1	17.8-18.8	-7.28	21-22	-4.04
MCV (fL)	80-81	63.5-65.1	-16.95	69.5-70.5	-11.52
Mean Corpuscular Haemoglobin Concentration (MCHC) (g/dL)	30.5-31.6	27.7-28.9	-2.74	30.3-31.5	-0.25
RDW (%)	14.1-15.1	16.3-17.3	2.17	27.3-28.3	13.19

Platelet count (PLT) (10^3/µL)	296.1-299.1	325-327	28.09	240-242	-57.41
HbF (%)	3.1-3.3	2.1-2.3	-1.03	51.9-53.1	49.66
HbA_2 (%)	2.3-2.6	5.1-5.4	2.75	2.4-2.7	0.08
HbA (%)	91.5-92.8	91.1-92.6	-0.15	42-45	-48.18

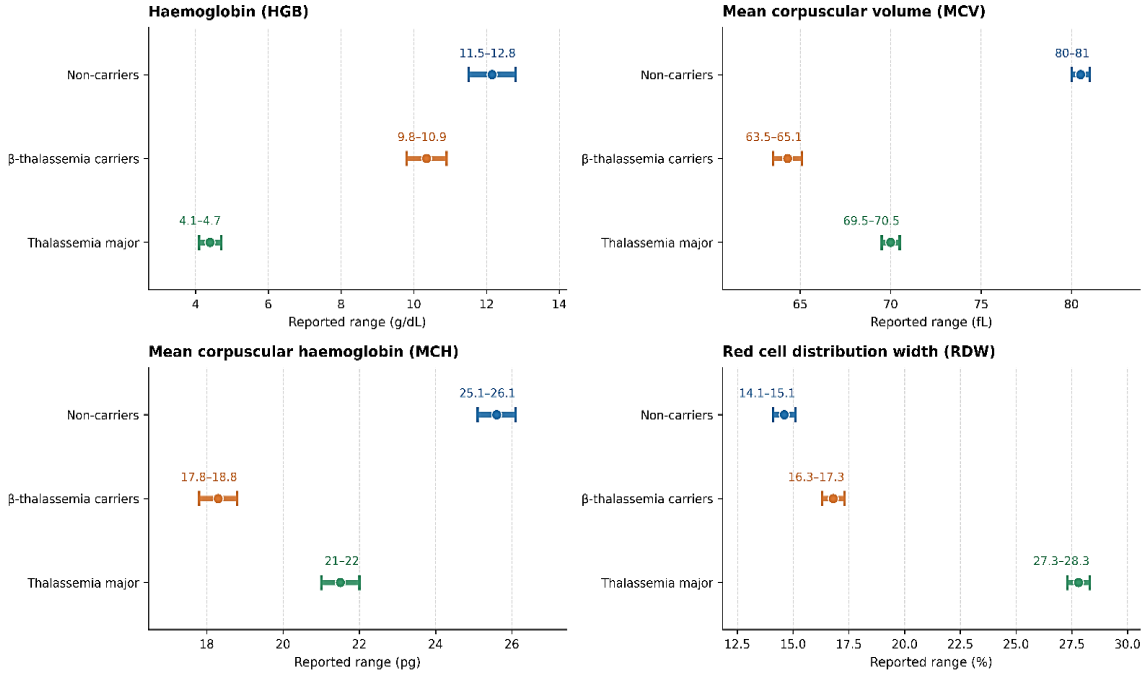


Figure 3: Comparison of haematological profiles between diagnostic groups

The clinical recommendations recorded included family screening and genetic counselling (27/85; 31.76%) and serum ferritin testing (27/85; 31.76%). Other recommendations included haematinic supplementation for six months (13/85; 15.29%) and repeat sampling after transfusion (1/85; 1.18%), as shown in Table 4.

Table 4: Clinical recommendations in diagnostic reports

Clinical recommendation	Counted	Percentage
Family screening and genetic counselling; rule out concomitant iron deficiency	27	31.76%
Serum ferritin testing	27	31.76%
Parents' carrier screening; clinical correlation; rule out concomitant iron deficiency	2	2.35%
Clinical correlation required	8	9.41%
Repeat sample after transfusion	1	1.18%
Haematinic supplementation for six months	13	15.29%
Consultation required	2	2.35%
Repeat CBC after iron profile and treatment, if needed	5	5.88%
Total recommendation entries counted	*85 (100%)*	

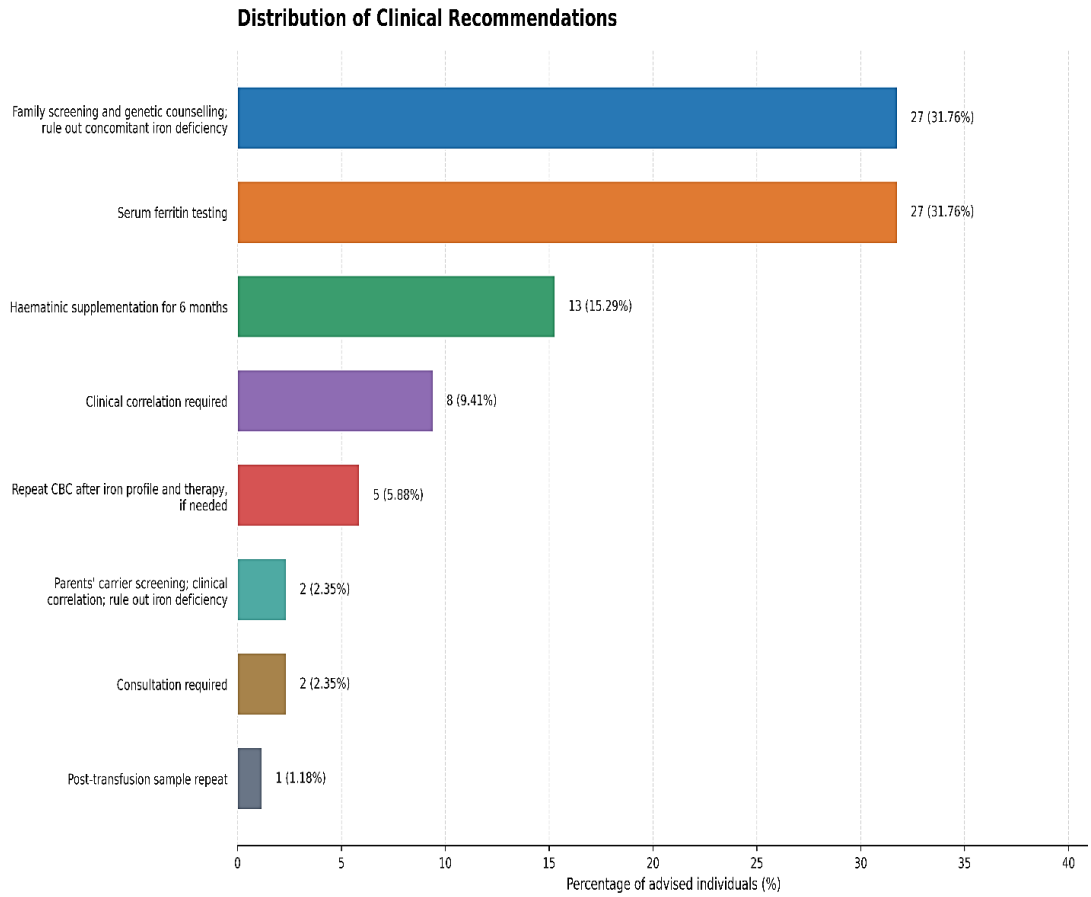


Figure 4: Distribution of clinical recommendations recorded in the 85 entries summarized following β-thalassemia screening in Narowal, Punjab, Pakistan. Bars represent the count and percentage assigned to each recommendation category.

DISCUSSION

This study aimed to describe the frequency of β-thalassemia and haematological profiles among individuals screened in Narowal, Pakistan. Of the 565 records reviewed, 21.59% were classified as β-thalassemia carriers. This figure is higher than some published carrier-frequency estimates, but direct comparisons are limited by differences in study populations and sampling methods. Ansari et al. (2012) reported that eight common β-thalassemia mutations accounted for 93.9% of the β-thalassemia alleles examined, with IVS1-5 being the most frequent mutation (40.89%). These findings describe mutation distribution and should not be interpreted as carrier prevalence in the general population.

Published estimates of β-thalassemia carrier frequency include 4.1% in Bangladesh (Khan et al., 2005) and 9.8% in Iran (Mirbehbahani et al., 2010). These estimates are not directly comparable with the 21.59% observed in this screened sample because the populations and sampling methods may differ. Although consanguinity is a relevant risk factor for autosomal recessive disorders, it was not measured in this study, so the observed carrier frequency cannot be attributed to consanguinity. Studies of affected families in Iraq and other settings provide context for the role of consanguinity, but do not establish a local causal association in Narowal (Ansari et al., 2012; Kadhim et al., 2017).

The frequency of thalassemia major in this screened sample was 0.35%. It should not be directly compared with reports of severely affected foetuses from prenatal diagnostic samples, because the populations and denominators differ. The low frequency observed here could reflect screening-selection effects, underdiagnosis, or the effects of preventive strategies; this retrospective dataset cannot distinguish among these explanations. Broader prenatal and postnatal surveillance and representative community sampling are needed to characterize the disease burden. These results alone do not establish Narowal as a genetic hotspot.

The haematological profile of the 122 β-thalassemia carriers showed an MCV of 64.97 fL, an MCH of 18.76 pg, and an HbA2 level of 5.32%. These findings are consistent with commonly used screening indicators for β-thalassemia trait, including MCV below 78 fL and HbA2 above 3.5% (Koyuncu & Aslan, 2001), and with comparable diagnostic thresholds reported in India (Madan et al., 2010). The observed microcytosis and hypochromia may also be influenced by concomitant iron deficiency or other factors; however, iron deficiency and HBB variants were not directly assessed in this study. These findings support the use of CBC and haemoglobin analysis in screening and the importance of appropriate iron studies when clinically indicated.

Among the 85 entries summarized in Table 4, 27 (31.76%) were recorded under family screening and genetic counselling. The available table does not establish how many of all 122 confirmed carriers actually received counselling, so counselling coverage among carriers cannot be calculated from these data. This distinction should be considered when assessing gaps in prevention services. Premarital screening programmes in other countries, including Kuwait, have reported identifying high-risk couples, but differences in programme design and population coverage should be considered when comparing outcomes (Rouh Aldeen et al., 2021). The findings support strengthening accessible carrier detection, counselling, and follow-up in Pakistan.

The study also highlights the potential value of molecular testing in Narowal. Ansari et al. (2012) reported that eight common β-thalassemia mutations accounted for 93.9% of the β-thalassemia alleles in their study, with IVS1-5 among the most common variants. These findings support consideration of regionally validated mutation panels; however, the present study did not perform molecular testing and therefore cannot establish which variants predominate in Narowal.

Research in China supports the value of characterizing region-specific thalassemia mutations to inform screening and counselling (Zeng et al., 2022). If future representative studies confirm a high carrier frequency in Narowal, a locally validated mutation panel could help guide efficient testing and counselling. A regional mutation registry could also support further research, awareness campaigns, and evidence-informed prevention policies.

This study included 565 screening records, which limits generalizability; sex-disaggregated data were not available. Further studies should include representative samples from adjoining districts, assess sex-specific trends, and analyse relevant inherited HBB variants, including IVS1-5, to inform regional screening strategies. The present findings may help guide the design of future screening and counselling services, but broader policy conclusions require confirmatory population-based evidence.

Conclusion and Recommendations

The present study identified a β-thalassemia carrier frequency of 21.59% among 565 individuals screened at DHQ Hospital, Narowal, with carriers exhibiting reduced MCV and MCH levels and elevated HbA2 compared with non-carriers. These findings highlight the importance of accessible screening and early identification; however, the results cannot be generalized to the entire district population because the study involved screening attendees. It is recommended that premarital and family-based screening, genetic counselling, community awareness, and appropriate hematological follow-up be strengthened. Serum ferritin testing should be considered when clinically indicated to assess possible iron deficiency. Future studies should use representative community samples, include neighboring districts, and incorporate molecular analysis of β-thalassemia-associated variants to establish regional carrier frequencies and guide evidence-based prevention strategies.

CONFLICT OF INTEREST

The authors declare no conflicts of interest.

FUNDING

This research received no specific funding.

AUTHOR'S CONTRIBUTION

Dr. Muhammad Asim identified the research gap and Mehwish Ishaq wrote the first draft of the manuscript. Ali Abbas performed the data collection. Abdul Qadoos and Muhammad Irfan analyzed the data and made the graphical work. Prof. Dr. Abdul Majid Khan and Prof. Dr. Muhammad Akbar Khan revised the manuscript.

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