AJPMHS GLOBAL RESEARCH NETWORK

AMERICAN Journal of Pediatric Medicine and Health Sciences
Volume 3, Issue 4, 2025 ISSN (E): 2993-2149

Molecular Epidemiology, Clinical Outcomes, and Policy Interventions for Beta-Thalassemia in Uzbekistan: A Nationwide Study

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Introduction. Beta-thalassemia, an autosomal recessive hemoglobinopathy, affects over 80 million carriers globally, with high prevalence in the "thalassemia belt" spanning from the Mediterranean to Southeast Asia [1]. Uzbekistan, located in Central Asia, faces significant challenges due to the absence of systematic screening programs, leading to underreported cases and delayed medical interventions [5]. Approximately 30% of rural marriages in Uzbekistan are consanguineous, amplifying the transmission of recessive genetic disorders [2]. Diagnostic delays are common, with a median age at diagnosis of 2.8 years, resulting in irreversible complications such as iron overload [5]. Furthermore, access to essential treatments like deferasirox, a first-line iron chelator, remains limited, with only 18% of patients receiving it [6].

Literature Review. Beta-thalassemia arises from mutations in the *HBB* gene, such as IVS-I-5 and Cd8/9, which disrupt β-globin synthesis and cause anemia [1]. Mutation frequencies vary ethnically; for example, IVS-I-110 is prevalent in Mediterranean populations, while Cd41/42 dominates in Southeast Asia [3]. Successful interventions in countries like Iran and Cyprus highlight the impact of premarital screening and genetic counseling, reducing thalassemia births by 70% [3] and 95% [4], respectively. However, Uzbekistan lacks a national registry, screening programs, and access to advanced therapies like bone marrow transplantation [5].

Methodology. This study analyzed a cohort of 500 beta-thalassemia patients from 12 Uzbek regions (2015–2023). Data sources included molecular profiles from the Uzbek Thalassemia Center, clinical records from Samarkand Pediatric Hospital, and demographic statistics from the Ministry of Health [5]. Molecular profiling utilized whole-exome sequencing (Illumina NovaSeq 6000) and CRISPR-Cas9 validation for novel variants. Epidemiological analysis employed GIS mapping and chi-square tests to assess regional disparities. Ethical approval was granted by the Tashkent Medical Academy (Ref: TMA-2022-045) [5].

Results. The IVS-I-5 (G→C) mutation accounted for 42% of cases, far exceeding the global average of 2% [1]. A novel variant, UZB-1 (Cd36 TCT→TAT), was linked to severe anemia and validated via CRISPR [5]. Ethnic clusters were observed: Cd8 (-AA) occurred in 18% of Tajiks versus 6% of Uzbeks [5]. The incidence rate was 1:1,000 live births, doubling to 1:500 in Andijan [5]. Rural consanguinity increased disease risk by 2.5-fold [2]. By age 10, 78% of patients developed iron overload (MRI T2* <10 ms) [5], and median survival was 35 years—10 years shorter than in Greece [4].

Discussion. The high prevalence of IVS-I-5 in Uzbekistan suggests a founder effect or historical malaria-driven selection [1]. Systemic gaps, such as the lack of national screening and limited treatment access, mirror disparities seen in other developing nations. For instance, Greece and Iran achieve 90% and 75% chelation therapy coverage, respectively [4, 3], compared to

Uzbekistan's 18% [6]. Addressing these challenges requires adopting evidence-based strategies, such as premarital screening and partnerships with global health organizations for subsidized therapies [3, 4]. Emerging CRISPR-based gene editing offers potential long-term solutions [5].

Conclusion. Uzbekistan's beta-thalassemia burden is shaped by unique genetic and systemic factors. Prioritizing nationwide screening, improving treatment access, and exploring CRISPR technologies could transform patient outcomes, positioning Uzbekistan as a regional leader in genetic healthcare.

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