HEMOCHROMATOSIS IN UZBEKISTAN: PREVALENCE, PROGNOSIS, AND CURRENT APPROACHES

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Abstract: Hemochromatosis is a genetic disorder characterized by excessive iron absorption and deposition in vital organs, leading to chronic complications such as liver cirrhosis, diabetes, cardiomyopathy, and arthritis. Although common in Western populations, especially those of Northern European descent, the condition is considered rare in Central Asia, including Uzbekistan. This article explores the current knowledge on the prevalence of hemochromatosis in Uzbekistan, the challenges in diagnosis, and what steps are being taken within the healthcare system to address this condition.

Introduction

Hereditary hemochromatosis (HH) is a metabolic disorder caused by mutations in genes involved in iron regulation, most notably the HFE gene (C282Y and H63D mutations). It leads to progressive iron overload if untreated. In high-

risk populations, HH occurs in approximately 1 in 200 to 1 in 400 individuals, but its prevalence is considerably lower in Asian populations.

Global and Regional Prevalence

- In Europe and North America, HFE-related HH is relatively common.
- In contrast, in Asian and Central Asian populations, including Uzbekistan, studies show that C282Y homozygosity is extremely rare or absent.
- A Turkish population study reported C282Y homozygosity at 0.043%, suggesting low prevalence in Turkic nations, which may include genetic similarities with Uzbeks.

Current State in Uzbekistan

- There are no large-scale epidemiological studies specifically focused on HH in Uzbekistan.
- Most hematological efforts are currently focused on iron deficiency anemia, which is far more common.
- Routine screening for HH or genetic testing (HFE, HJV, HAMP, TFR2) is not widely implemented.
- Cases of hemochromatosis may go underdiagnosed or misdiagnosed, particularly in older adults with liver, joint, or cardiac symptoms.

Diagnosis and Prognosis

- Early diagnosis is key to preventing irreversible organ damage.
- Diagnostic criteria often include:
    - Elevated serum ferritin
    - Increased transferrin saturation
    - Genetic testing for HFE mutations (C282Y and H63D)

- In Uzbekistan, phlebotomy is available as a standard treatment if iron overload is confirmed.
- Prognosis is excellent if treated early, but late-stage disease carries risk for cirrhosis, hepatocellular carcinoma, and cardiac failure.

Treatment and Management in Uzbekistan

- Phlebotomy (therapeutic blood removal) is the primary treatment for HH and is available in major medical centers.
- Iron chelation and dietary management are used in selected patients.
- However, awareness among physicians is low, and no dedicated HH programs or registries currently exist.
- There is a need for better diagnostic protocols and integration of genetic testing into national labs or partnerships with international labs.

Challenges

- Low awareness among the general public and healthcare providers.
- Limited access to genetic testing and advanced diagnostics.
- Absence of nationwide data or clinical guidelines specific to hereditary iron overload.

Conclusion

Although hereditary hemochromatosis is likely very rare in Uzbekistan, the lack of dedicated research, awareness, and diagnostic infrastructure means some cases may remain undiagnosed or mismanaged. Given the availability of simple and effective treatment like phlebotomy, improving education among clinicians, implementing basic iron overload screening in high-risk patients, and introducing affordable genetic testing would be beneficial. Long-term, establishing a national

registry and conducting population-specific studies will be crucial to understanding the true burden of the disease in Uzbekistan.

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