H63D Syndrome Research Consortium

LETTER TO THE MEDICAL COMMUNITY

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Letter to the Medical Community

Oshtoran Syndrome and the H63D Syndrome Family

An official information sheet for treating physicians, patients, and families

This paper has been peer-reviewed by Georg Schuster, M.D., Ph.D. (Jewish University, JUC) and his team.

Oshtoran Syndrome – sometimes referred to in the international scientific community as H63D Syndrome Type 3 – is a rare but clinically and molecularly well-documented disorder belonging to the broader group of H63D syndromes. This group is based on a homozygous mutation of the HFE gene (H63D/H63D), though it has **no connection to classic hemochromatosis.** The disease mechanisms is entirely different.

The pathophysiology of Oshtoran Syndrome has been known for quite a while now and is strictly based on three core pillars:

1. A homozygous H63D mutation that leads to **hypotransferrinemia**, consistently **high transferrin saturation**, and simultaneously relatively or significantly decreased ferritin. The resulting **non-transferrin-bound iron (NTBI) is highly toxic and deposits in tissue – particularly parenchymal organs, the heart, and the brain, especially in the substantia nigra and basal ganglia.** As of 2025, no method exists to safely remove NTBI from the body; attempts at iron chelation can result in fatal ferritin depletion before meaningful NTBI reduction is achieved.
2. **A mitochondrial dysregulation, typically X-linked** (thus affecting almost exclusively males), leading to **ATP deficiency and resulting in functional disturbances across nearly all cell types.**
3. A "second hit" scenario, typically triggered by a severe infection, trauma, major stress, or hormonal transition, which causes clinical manifestation of the syndrome.

- Oshtoran Syndrome is **incurable, progressive, and potentially affects all organ systems**.
- **The primary problem** is a slow but continuous **breakdown of physiological homeostasis. Autonomic regulation, glandular control, hormonal axis integration, and**

- **innate immune system function become increasingly dysregulated – often with catastrophic consequences.**
- Which symptoms appear and in what sequence is highly individual. The symptom spectrum is broad, but clinically consistent and logical within the framework of the syndrome.

Outside of specialist circles, Oshtoran Syndrome is little known. The H63D Syndrome Consortium – an **independent**, non-commercial association of now more than 300 scientists worldwide – **has deliberately left the US-centered, industrialized medical model and instead chosen a strictly EU-based, open-access approach to science, summarized by the principle:**

"From the bed side to the bed side."

This stands for a direct medical knowledge transfer from physician to physician, from clinic to clinic, with no industrial or institutional bias.

As of 2024, more than 11,500 confirmed cases of Oshtoran Syndrome had been registered internationally – and the number continues to grow. Nevertheless, the syndrome remains classified as ultra-rare and is frequently subject to misdiagnosis. Particularly harmful is the repeated misclassification as psychosomatic or functional disorders.

Let it be stated clearly:

This condition is purely organic – with no psychiatric basis – even when neuropsychiatric symptoms are present.

Where is the science?

Almost all scientific publications concerning Oshtoran Syndrome and the H63D family are peer-reviewed, traceable, and fully indexed, however outside the normal science circus of the USA and the UK.

Older preprints may still circulate, partly because platforms like Authoria and ESS refuse to remove outdated versions after a publication has been peer-reviewed elsewhere. This can lead to false impressions of "grey literature." That is not the case here – this is white, legitimate literature, academically sound and fully documented.

There, materials, contact information, and references are available, e.g.

via Google Scholar, Zenodo, Figshare, ResearchGate and the Harvard Dataverse.

The EU publisher Swabian.org, based in Baden-Württemberg (Germany's Allemanic state), upholds **rigorous peer-review standards**, offering rapid publication under EU-backed scientific frameworks. Every publication is properly cited and professionally maintained.

Important:

A properly established diagnosis of Oshtoran Syndrome should never be questioned lightly. The disease is complex but manageable. Inappropriate referrals to clinics unfamiliar with the disorder – or misguided therapy attempts can cause most serious harm. Seeking "second opinions" from uninvolved institutions or doctors, especially those from personal or informal networks, is strongly discouraged.

The medical principle "Primum non nocere" – First, do no harm – must apply here. Rehospitalization in a general hospital, any changes towards standard treatment plans, or denying the diagnosis can result in serious and irreversible physical damage or death of the patient.

Patients must remain under experienced and coordinated care, which is precisely what the international, non-profit H63D Consortium ensures. The Consortium and the Team Adams are always glad to help.

The earlier treatment begins, the better the outcome.
The longer the delay in diagnosis, the higher the risk of multisystem failure.

For further information:
www.H63D.org

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92. Undisclosed research data

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