HFE H63D mutation: why are you killing your patients?

Dr. Carolina Diamandis[1], Marius Lazar[2], Lucas Smith[2], Olga Ivanova[2], David Seideman[2,3], Marianne Kaufmann[4], Sophia Papadopoulou[2], Adrian Tudor[2], Jonathan Feldman[2], Georg Schuster[3], Riku Honda[4], and Jacob Adams[4]

[1]Affiliation not available
[2]LCG Greece Research
[3]Jewish University of Colorado
[4]International H63D Syndrome Research Consortium

May 16, 2022

Abstract

Evidence-based medicine has shown for many years that homozygous mutations of the HFE gene H63D are by no means negligible. Not only can it cause, usually after a second hit, rather mild classical hemochromatosis, but it can also cause numerous other disorders of iron metabolism, such as hypotransferrinemia, changes in binding capacity, and others. In addition, it may lead - among other symptoms - to damages of the heart and the substantia nigra via a causal relationship that remains to be investigated, most likely via a cascade dysfunction in iron metabolism. The clinical facts are compelling. Any physician who dismisses mutations of the HFE gene H63D as clinically irrelevant risks the health and life of his patient. We report two patients with an HFE H63D mutation who were treated almost too decades too late because of outdated expertise.

Hosted file

`ignoring h63d is fatal.pdf` available at https://authorea.com/users/410930/articles/569361-hfe-h63d-mutation-why-are-you-killing-your-patients