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ABSTRACT

Level Of Awareness among Pediatricians in Palestine about Urea Cycle Disorders with A Scope on HHH Syndrome.

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Urea cycle disorders (UCDs) are inborn errors of nitrogen detoxification and arginine synthesis caused by defects in urea cycle enzymes. Most UCDs are autosomal recessive, increasing the likelihood of occurrence in the Palestinian population due to the high rate of consanguineous marriages. These disorders primarily affect liver and brain function. One of the rarest among them is Hyperornithinemia–Hyperammonemia–Homocitrullinuria (HHH) syndrome, an autosomal recessive condition resulting from mutations in the SLC25A15 (ORNT1) gene, which disrupts the mitochondrial ornithine transporter. Fewer than 100 cases have been reported globally.

HHH syndrome presents with a broad spectrum of neurological symptoms, including intellectual disability, hypotonia, spasticity, convulsions, and cerebellar ataxia, as well as acute metabolic decompensation episodes such as coma, liver dysfunction with coagulopathy, and developmental delays. As with other UCDs, protein intake management is essential to reduce ammonia buildup and prevent serious complications.

This study utilized a survey-based approach to assess the level of awareness among at least 50 Palestinian pediatricians. The questionnaire covered general knowledge of UCDs and

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progressively focused on HHH syndrome. It addressed awareness, education, diagnostic approaches, complications, and clinical experience, targeting both general pediatricians and those with a metabolic focus.

The results revealed a significant gap in awareness and knowledge of UCDs among Palestinian pediatricians. While approximately 70% acknowledged the genetic nature of these disorders, many lacked detailed understanding—particularly regarding HHH syndrome and key biochemical markers like hyperammonemia. Over half had never attended training related to metabolic or genetic diseases, and 32% reported limited access to advanced diagnostic tools. Furthermore, only a minority expressed confidence in recognizing or diagnosing HHH syndrome, despite 80% linking consanguinity to the prevalence of such disorders. Encouragingly, most respondents recognized the importance of early diagnosis and supported increased access to workshops and professional development.

This research aims to improve patient outcomes in Palestine by addressing the severe consequences of delayed diagnosis and enhancing knowledge exchange between general pediatricians and specialists. It also underscores the urgency of updating medical education, providing better diagnostic resources, and implementing structured training sessions focused on metabolic and genetic diseases. Moreover, the study highlights the importance of equipping healthcare professionals with the critical thinking skills needed for handling rare disorders like HHH syndrome. Ultimately, this work encourages further research and strategic planning to close knowledge gaps and promote better clinical readiness in the Palestinian healthcare system.