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LETTER TO THE EDITOR

Reply—Nonimmune Hydrops Fetalis and Lysosomal Storage Diseases



To the Editor,

We thank the author for his comments focusing on the wide range of incidence (1-18%) of nonimmune hydrops fetalis (NIHF) due to inherited metabolic diseases, including lysosomal storage diseases (LSDs). This wide range of incidence may arise from many reasons as discussed below.

First of all, in developed western countries, the low incidence of inherited metabolic diseases because of infrequent consanguineous marriages is expected to be the real cause of low incidences. Additionally, in developed countries, the low index of clinical suspicion because of the inadequate clinical experience of physicians may cause a pseudo-reduction in the incidence. However, in developing countries, the low index of clinical suspicion or lack of appropriate diagnostic laboratory technologies, and financial and technical problems in sending biological specimens of the cases to referral centers in developed countries and getting the diagnoses may cause a pseudo-reduction in the incidence. More importantly, severe cases may be lost by abortions or early intrauterine deaths during the prenatal period, with the result that they could not receive detailed investigation and diagnosis. All these factors could contribute to low incidence of NIHF due to LSDs.

On the other hand, high incidence of NIHF due to LSDs could be caused by the high incidence of consanguineous marriages and inherited metabolic diseases in developing countries. However, in developed countries, owing to referral centers receiving patients and biological specimens from all over the world, the incidence is expected to be high. Therefore, it is very difficult to calculate the real incidence of such rare inherited metabolic diseases globally, and a high index of clinical suspicion supported by appropriate laboratory investigations to establish the diagnosis should be more important than calculating the incidence.

Ozge Surmeli-Onay* Ayse Korkmaz Section of Neonatology, Department of Pediatrics, Hacettepe University Faculty of Medicine, Ankara, Turkey

*Corresponding author. Hacettepe University Ihsan Dogramaci Childrens' Hospital, Neonatology Unit, 06100, Ankara, Turkey. *E-mail address:* ozgeonay79@gmail.com (O. Surmeli-Onay)

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