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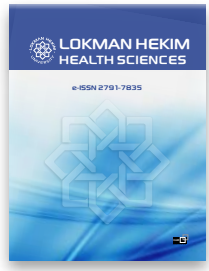

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LETTER TO THE EDITOR

Spinal Muscular Atrophy Screening in Türkiye: From Awareness to Clinical Outcomes

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Dear Editor,

I read with great interest the article entitled "Awareness and Attitudes Toward Spinal Muscular Atrophy (SMA) Carrier and Newborn Screening Programs in Türkiye: A Cross-Sectional Study in Family Health Centers" by Özbaş et al.[1] The authors should be commended for providing valuable data on a recently implemented nationwide screening initiative.

The study clearly demonstrates that information and awareness play a significant role in individuals' willingness to participate in SMA screening. These findings are of great importance, particularly for Türkiye, where both premarital carrier screening and newborn screening have recently been integrated into national health policies. As the authors emphasize, increasing public awareness is essential to improve the effectiveness of these programs.

In recent years, the clinical management of SMA has changed significantly. The use of disease-modifying therapies such as nusinersen, onasemnogene abeparvovec, and risdiplam has improved the natural course of the disease.[2–4] Studies have shown that starting treatment before symptoms appear leads to much better motor and survival outcomes.[2–4] Recent studies also show that newborn screening enables early diagnosis and timely treatment, which is directly associated with better clinical outcomes.[3] Therefore, SMA screening programs should be considered not only as a tool for early diagnosis but also as a crucial step in enabling early and life-changing treatments.

In our country, nusinersen is widely used and remains the primary treatment option under the national reimbursement system.[2] Although new treatment options such as risdiplam have entered clinical use, access is limited by certain criteria. Onasemnogene abeparvovec gene therapy is available only in limited settings due to cost and restrictions related to health policies. These factors further increase the importance of screening programs for early diagnosis and timely treatment. This is particularly relevant, as recent evidence shows that early diagnosis and timely treatment can significantly modify the course of the disease and long-term clinical outcomes.[4,5]

Considering the relatively high carrier frequency and the nationwide implementation of screening programs, increasing public awareness in Türkiye is especially important. However, it is also important that individuals not only access screening but also understand its clinical value.

In this context, the role of primary care physicians and structured genetic counseling is very important. Open and effective communication at the primary care level may help translate awareness into actual participation in screening programs. However, it is still unclear how awareness translates into real screening uptake, early treatment, and

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long-term neurological outcomes. Future studies focusing on these points would provide valuable information about the real-world impact of screening programs.

In conclusion, this study provides important insight into public awareness of SMA screening in Türkiye. In the current treatment era, increasing awareness is not only a public health goal but also a key factor for early diagnosis and effective treatment.

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