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Study title: MAGT1 Deficiency: Detailed Clinical and Laboratory Features with Treatment Options and Outcomes

Dear ESID members,

MAGT1 deficiency, also known as XMEN syndrome, is an X-linked condition caused by hemizygous mutations in the MAGT1 gene (1). It is classified as an inborn error of immunity, with fewer than 70 patients described in the literature to date. Due to its rarity and the limited number of reported cases, the full clinical spectrum and outcome of the disease has not yet been clearly defined (2).

Since its first description in 2011, significant advances have been made regarding the genetic etiology, immunopathology, and clinical features of this syndrome. However, there is still a lack of comprehensive data on the broader clinical manifestations and therapeutic approaches. In addition, only a limited number of patients undergoing HSCT have been reported in the literature, and post-transplant bleeding-related complications have been documented. Evaluating these transplanted patients and assessing their survival is also one of our objectives.

As the Department of Pediatric Immunology and Allergy at Marmara University School of Medicine in Türkiye, we are initiating a collaborative study on patients with MAGT1 deficiency. Our aim is to comprehensively characterize their clinical presentations, immunological profiles, genetic findings, and treatment outcomes. So far, data from 32 patients have been collected. Expanding this cohort with additional cases will significantly enhance our understanding of XMEN disease and contribute to the development of standardized care protocols.

We invite ESID centers and members who have cared for patients with MAGT1 mutations to participate. Data collection will be performed via an Excel-based CRF that will be shared upon expression of interest. If you have one or more patients with genetically confirmed MAGT1 deficiency, we would be very grateful for your participation. Please contact us directly and we will provide the CRF and further instructions.

Your participation would be a great honor for us, and we believe that, together, we can advance our understanding of this rare disorder.

With best wishes,

On behalf of the MAGT1 Study

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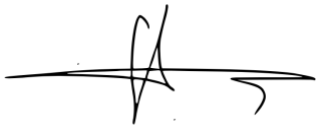

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References

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