

How Early Diagnosis can be Improved in Primary Immunodeficiency (PID): The First Italian-Turkish Joint Workshop

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Dear Editor,

Effective communication among medical professionals can facilitate the diagnosis, treatment, and care of individuals with primary immunodeficiency (PID). In November 2023, the first Italian-Turkish joint workshop on PID was held at a tertiary academic campus, with 50 physicians, including pediatric and adult immunology-allergy subspecialty residents and specialists.

The one-day workshop, conducted in collaboration between the Immunology Department of Anna Meyer Children's Hospital at Florence University and the Department of Pediatric Immunology-Allergy at Ankara University, aimed to discuss current challenges in PID diagnosis and to raise awareness of newborn screening (NBS) for severe combined immunodeficiency (SCID) among pediatric and adult clinical immunologists in both countries. Early

diagnosis of SCID, a pediatric emergency, is essential for survival and better outcomes by enabling early treatment (1,2). Infants with SCID meet the criteria for NBS because they have no symptoms at birth, the disease is fatal if left untreated, and the prognosis is much better with early treatment. Therefore, NBS is vital for infants with SCID (3). A recent study by Haskoloğlu et al. highlighted the importance and outcome of NBS in Türkiye (4).

The workshop focused on the current status of PID diagnosis, barriers to early detection, and educational and research opportunities to improve timely diagnosis.

Four sessions addressed the current state of diagnosis in pediatric and adult PID, newborn screening for SCID, illustrative pediatric and adult cases in which diagnosis had been delayed, and interactive discussions on practical needs and next steps.

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The discussions highlighted several shared barriers to early diagnosis, including delayed referral, limited access to advanced diagnostic testing, incomplete recognition of warning signs, and gaps in continuity of care during transition from pediatric to adult services.

Participants proposed educational and organizational strategies to address these barriers, including structured training for pediatric and adult clinicians, summer schools and rotation programs for subspecialty residents, regular online case discussions, practical guidance on advanced diagnostic tests and treatment approaches, and closer collaboration among referral centers, health authorities, patient organizations, and patient associations.

In conclusion, this first Italian-Turkish joint workshop emphasized the need to strengthen communication and collaboration among physicians involved in the diagnosis and treatment of PID in both countries. The agreed initiatives include expanding education and research opportunities, preparing practical guidance for advanced diagnosis and treatment, and developing comprehensive transition protocols for patients moving from pediatric immunology care to adult services. These protocols should include coordinated joint visits, shared transfer summaries, and psychosocial support to facilitate continuity of care as patients reach adulthood.

Conflict of Interest

The authors have no conflicts of interest to declare.

Funding

The first Italian-Turkish joint workshop on PID was supported by Kedrion Biopharma.

Author Contributions

Concept: **AI**, Design: **AI**, Data collection or processing: **AI**, **NDD**, Analysis or Interpretation: **AI**, **NDD**, Literature search: **AI**, Writing: **AI**, **NDD**, Approval: **All authors**.

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