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Title of Presentation	Hereditary Alpha-Tryptasemia in Mastocytosis: A Turkish Single-Center Experience Using Digital PCR-Based TPSAB1 Copy Number Analysis
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Abstract of Presentation	Background: Hereditary alpha-tryptasemia (HαT), caused by increased germline TPSAB1 alpha-tryptase copy number, is increasingly recognized as a modifier of mastocytosis. However, data from Türkiye are lacking. We evaluated the frequency and clinical impact of HαT in a well-characterized mastocytosis cohort using digital PCR (dPCR). Methods: TPSAB1 copy number variation was analyzed in peripheral blood genomic DNA from 124 patients with mastocytosis and 71 healthy controls using the QuantStudio Absolute Q dPCR system. HαT was defined according to established copy number criteria. Clinical, laboratory, and molecular characteristics were compared between HαT-positive and HαT-negative patients. Results: TPSAB1 status was evaluable in 121 patients. HαT was identified in 7 patients (5.8%) and was absent in healthy controls. HαT-positive patients had higher baseline serum tryptase (median 68 vs 24.8 µg/L; p=0.014). Gastrointestinal involvement was more frequent (71.4% vs 20.5%; p=0.002), whereas cutaneous involvement was less frequent (28.6% vs 76.1%; p=0.017). Anaphylaxis was more common in HαT-positive patients (57.1% vs 29.9%), although not statistically significant. Peripheral blood KIT D816V variant allele frequency was comparable between groups. HαT-positive

patients also had higher monocyte and basophil counts and lower serum IgM levels. Conclusions: HαT was detected in patients with mastocytosis but not in healthy controls, supporting its enrichment in mastocytosis. This study provides the first comprehensive evaluation of HαT in a Turkish mastocytosis cohort using dPCR-based TPSAB1 copy number analysis. Beyond elevated baseline serum tryptase, HαT was associated with a distinct clinical phenotype characterized by greater gastrointestinal involvement and reduced cutaneous disease. These findings highlight the clinical relevance of TPSAB1 copy number assessment and support its further evaluation in larger multicenter studies. Keywords: digital polymerase chain reaction; hereditary alpha-tryptasemia; KIT D816V; mastocytosis; TPSAB1; tryptase