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A case of an elevated tryptase

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Background

- Hereditary Alpha Tryptasemia (HαT) is autosomal dominant disorder characterized by elevated baseline tryptase (8 ng/ml or greater) and increased copies of α-tryptase alleles
- Occurs in 3% of the population and more common in female patients

Case presentation

- A full term, 13-month-old female patient presented to clinic for an elevated tryptase
- Past medical history: gastroesophageal reflux disease
- Past surgical history: none
- Family history: older sister with history of flushing. She eventually had a negative evaluation for mastocytosis
- Medications: none

Case presentation (cont.) and evaluation

	Presentation	6-mo F/U	9-mo F/U	12-mo F/U
Symptoms	Loose stools	Fussiness, increased night awakenings, and constipation (no loose stools)	Worsening fussiness, return of loose stools, first episode of facial flushing	Symptoms controlled
Tryptase (ng/ml)	13.0	11.7	12.2	
Other labs	CBC with thrombocytosis and eosinophilia and peripheral blood *c-kit* D816V negative	CBC normalized	Sent microarray for copy number variants of *TPSAB1, TPSB2, TSPG1, TPSD1*	Microarray showed extra allelic alpha-tryptase at *TPSAB1*
Attempted treatment	Cetirizine as needed + epinephrine autoinjector	Added cromolyn	Cromolyn dose adjusted due to side effects	Continue cetirizine and cromolyn

Management

- Diagnosis is made based on symptoms, serum tryptase level, and genetic testing
- Treatment is supportive in nature and includes maximally tolerated H1 and H2 histamine blockers as well as cromolyn for gastrointestinal symptoms
- In patients with HαT and known hypersensitivity, epinephrine autoinjector is recommended.

Conclusion

- Given a higher prevalence than systematic mastocytosis, one must have a high degree of suspicion for HαT to include in one's differential diagnoses
- Number of allelic copies of alpha tryptase correlates with symptom severity

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