A case of an elevated tryptase

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Background

Hereditary Alpha Tryptasemia (HαT) is an autosomal dominant disorder characterized by elevated baseline tryptase (8 ng/ml or greater) and increased copies of α-tryptase alleles. It occurs in 3% of the population and is 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.

Symptoms

- Presentation: Loose stools.
- 6-mo F/U: Fussiness, increased night awakenings, and constipation (no loose stools).
- 12-mo F/U: Symptoms controlled.

Tryptase (ng/ml)

- 6-mo F/U: 13.0
- 9-mo F/U: 11.7
- 12-mo F/U: 12.2

Other labs

- CBC with thrombocytosis and eosinophilia and peripheral blood c-kit D816V negative.
- CBC normalized.

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.

Conclusion

• 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.

References