COMPLEX, PERSISTENT, MULTI-GENERATIONAL PRESENTATION OF HEREDITARY ALPHA TRYPTASEMIA SYNDROME REQUIRING HIGH DOSE OMALIZUMAB

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Background: Hereditary Alpha Tryptasemia Syndrome (HATS) is a rare, complex disorder. The presentation can be intermittent or chronic and with local or multisystem involvement with a baseline tryptase level above >8-10 ng/ml. Genetic testing revealing additional copies of alpha-tryptase at TPSAB1 confirms the diagnosis.

Materials/Methods: Case History: Herein, we present a 67 yo Female with a lifelong history of urticaria, diagnosed with Chronic Spontaneous Urticaria (CSU) and mastocytosis. Additional medical history includes Ehlers-Danlos syndrome, postural orthostatic tachycardia syndrome, immunodeficiency, drug allergies, rheumatoid arthritis, asthma, chronic bronchitis, Hashimoto's thyroiditis, Sjogren's Syndrome, and Raynaud's Syndrome. The patient suffered lifelong chronic urticaria manifested by itching, hives, intermittent throat swelling with Urticaria Activity Score over Seven days (UAS7) score greater than 30 with Tryptase level of 13.8 ug/L. Genetic testing confirmed three copies of alpha-tryptase (additional copy at TPSAB1), two copies of beta-tryptase. Her two daughters (39-40yo) have similar presentations of urticaria and elevated tryptase.

Results: The Challenge in Treatment and management: The FDA approved dosage of omalizumab for CSU of 150 or 300 mg every 4 weeks was not adequate to provide symptoms control. However, omalizumab 300 mg every 2 weeks provided a remarkable reduction in symptoms to mild itching UAS7 (1-6). Tapering to every 3 or 4 weeks increased flare-ups.

Conclusions: We describe a multi-generational presentation of HATS requiring high dose omalizumab for adequate symptom control. When symptoms of CSU remain uncontrolled on omalizumab 300 mg q 4 weeks, more frequent dosing may be necessary.