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Two ladies with nC1-INH-HAE suffering from massive abdominal attacks: a diagnostic & therapeutic challenge

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Rationale: Hereditary Angioedema, HAE, with nC1INH is a very rare disease, and especially in sporadic cases, difficult to diagnose. Besides exclusion diagnosis, genetics are the only reliable diagnostic method if a known mutation results. Here we report two ladies with primarily abdominal attacks.

Methods: Patient 1) a 61-year-old lady suffered from recurrent massive painful abdominal swellings for many days. She was seen by gynecologists, pain specialists, surgeons and psychiatrist. Finally, she was sent to see the HAE specialist. Patient 2) a 32-year-old lady suffered from mild skin-swellings since she was a teenager without seeing any MD. Since 3 years, she also suffered from massive abdominal attacks and leg swellings not able to wear trousers.

Both ladies were checked for C1-INH-HAE, acquired angioedema, other reasons for swelling, all resulting negative. Genetic testing is pending. In both cases, we arranged appointments within an acute attack and treated with Icatibant SC.

Results: 30min after icatibant, both patients reported a striking symptom-relief and after 1h they were symptom-free. Thus the diagnosis nC1-INH-HAE was confirmed. Unfortunately, both ladies suffered from 1 to 2 attacks weekly. Patient 1) received firstly tranexamic acid resulting in attack number and severity reduction. As she did not achieve symptom-freeness, treatment was changed to oral beroltralstat. In patient 2) we firstly started with pdC1INH prophylaxis IV. As she still developed attacks, treatment was changed to lanadelumab.

Conclusion: Both patients are doing very well with their treatment that is in any case off-label for the indication right now. Data from: Austrian Registry of Bradykinin-mediated Angioedema, IIR-AUT- 002507.