



AAE with C1 inhibitor deficiency and ocular lymphoma: case report

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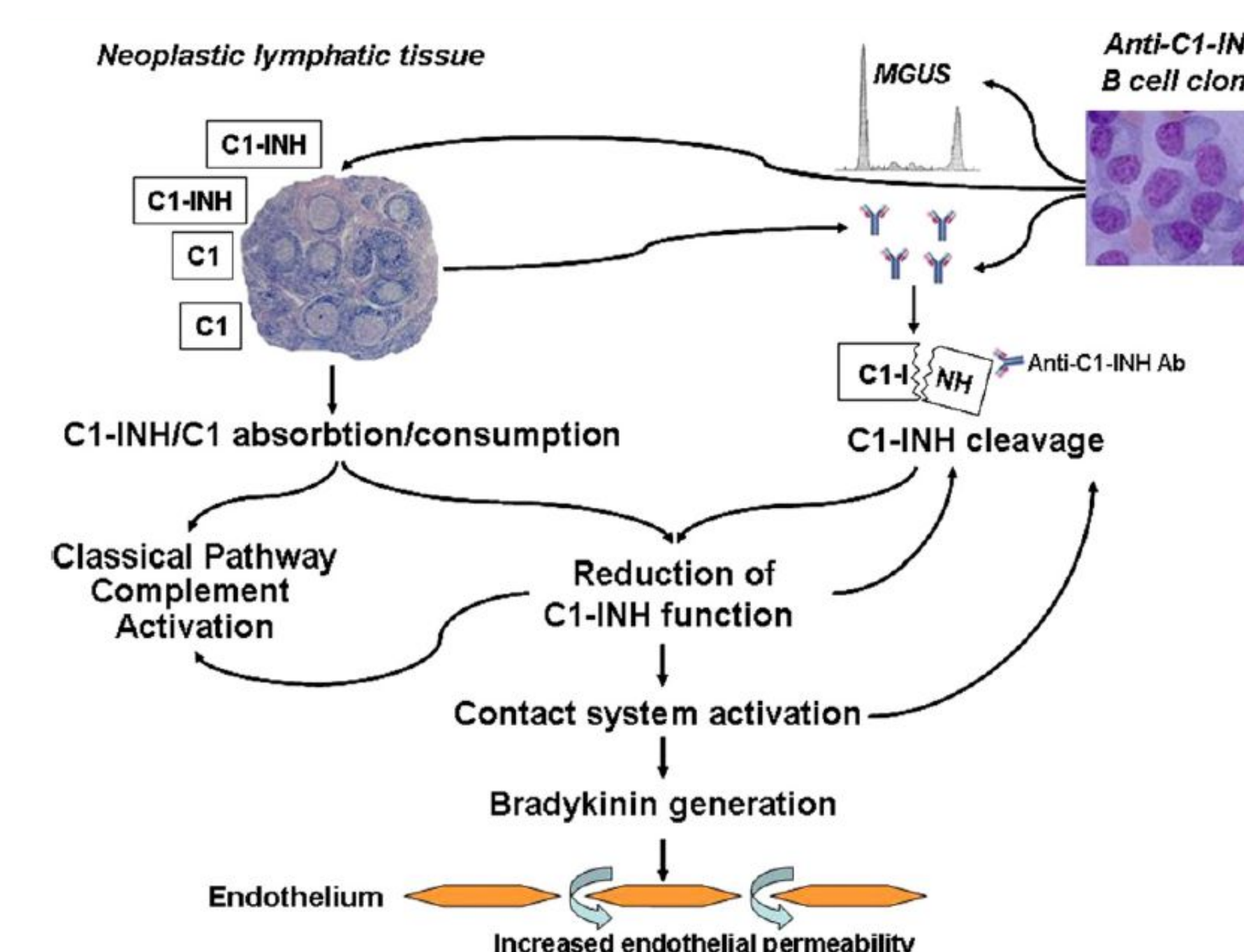
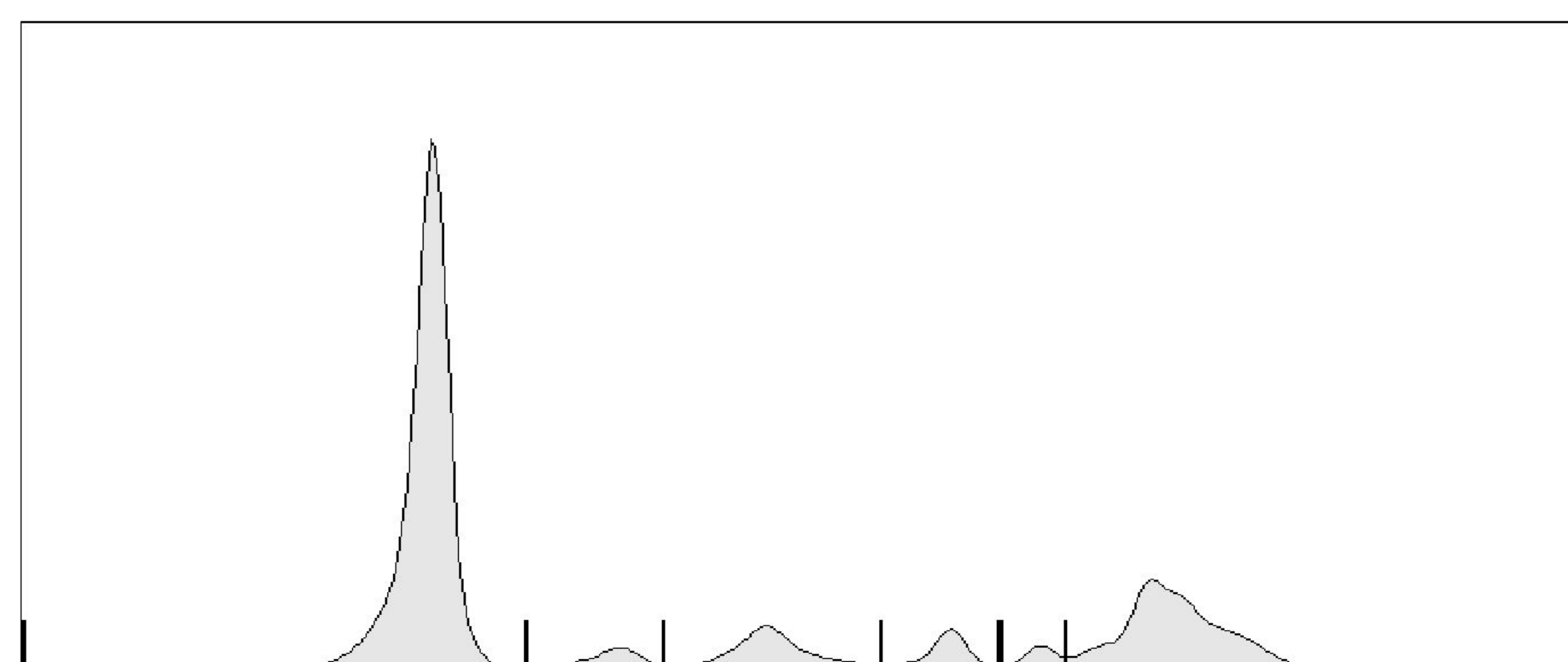
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BACKGROUND

AAE is a very rare condition, frequently associated with MGUS and in some cases with lymphoproliferative or autoimmune diseases. Its pathogenesis is related to the consumption of C1 inhibitor (C1-Inh) by neoplastic lymphatic tissue and/or the formation of neutralizing anti-C1Inh autoantibodies. Angioedema often comes before the onset of hematologic disease by years: in this case appears an ocular lymphoma after 25 years from AAE C1 inhibitor deficiency diagnosis.

METHODS

75 years old woman with diagnosis of AAE with C1 inhibitor deficiency secondary to MGUS IgA type k treated by icatibant. In March 2021 appeared ocular pain treated by ophtalmologist with topic steroids. The pain doesn't improve so the ophtalmogist made ultrasound that suggested an ocular lymphoma.



PICTURE 1. Ref. Cugno et al. Autoimmunity Reviews 2008; 8: 156

Protein fractions	Value	Reference values
Albumin	56,2%	55,8-66,1%
Alfa 1	3,9%	2,9-4,9%
Alfa 2	9,5%	7,1-11,8%
Beta 1	5,0%	4,7-7,2%
Beta 2	2,6%	3,2-6,5%
Gamma	22,8%*	11,1-18,8%
Ratio	1,28	1,10-2,40

Remarks: 1 MC IgA type kappa 7% and 1 MC IgM type lambda 5% in gamma fraction

Angioedema	C1 INH antigenic	C1 INH functional	C4	C1q	C1 INH antibodies
HAE type I	<50%	<50%	Low	Normal	Negative
HAE type II	>50%	<50%	Low	Normal	Negative
AAE	>50%/<50%	<50%	Low	Positive in 70%	Positive in 70%

RESULTS

The patient was hospitalized in Immunological Clinic in Naples where she performed hematochemical investigations showing new monoclonal component in lambda-type gamma IgM region. MRI illustrated hypointense tissue that included omolateral optic nerve compatible with lymphoid origin. PET-TC was negative for metastasis. Histological examination showed peripheral B lymphocyte-derived lymphoma compatible with marginal zone lymphoma. Therefore, she underwent radiotherapy from 17th March 2022 to 4th April 2022 with regression of lymphoid tissue at orbital MRI checkup in November 2022.

CONCLUSIONS

So it's very important perform hematological screening in patients with AAE not only at the time of diagnosis but also in the follow-up. Similarly, the hematologist should raise the suspicion of AAE in those cases of hematological disease of uncertain origin with typical manifestations.

