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Fatal case of ataxia telangiectasia complicated by severe epistaxis due to nasal telangiectasia in a 12 year old boy

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Summary.

Ataxia telangiectasia (AT) is a rare autosomal inherited disorder with progressive ataxia, cutaneous and ocular telangiectasia, and an increased risk for malignancies due to chromosomal instability and varying degrees of immunodeficiency.

Telangiectasia mostly occurs in eyes and sun exposed skin areas. However, mucosal bleeding due to telangiectasia within the vesical bladder has been reported. We, for the first time, report massive epistaxis in a 12 year old boy with AT due to multiple endonasal telangiectasias. Bleeding was refractory to a combined tamponade but could successfully be controlled using laser vaporization. Despite control of bleeding the boy -who had suffered

from progressive chronic liver disease of unknown origin- died from liver failure 15 days after laser treatment.

In conclusion, telangiectasias in mucous membranes have to be considered in AT patients with bleeding events. In particular, epistaxis in patients with AT should prompt for ENT examination to diagnose and treat endonasal telangiectasia.

To the editor.

Ataxia telangiectasia is a rare autosomal inherited disorder due to mutations in the *ATM* gene (Ataxia telangiectasia mutated) that affects 1 in 40.000 - 100.000 live births. Patients show progressive ataxia due to cerebellar degeneration as well as cutaneous and ocular telangiectasia. Due to chromosomal instability, there is an increased risk for malignancies. A combined immunodeficiency can occur affecting both B and T cells (reviewed in 1). Steatosis hepatis with increased liver enzymes appears to be common in patients with AT and may progress to advanced liver disease at young age (2).

Hematuria due to vesical telangiectasia has been described in AT patients undergoing chemotherapy (3-5), indicating that mucous membranes can be affected in patients with AT. Epistaxis due to endonasal telangiectasia has not been described previously in AT patients.

Patient history: A 12 year old boy with AT presented at our emergency department after prolonged epistaxis over six hours. AT had already been diagnosed at the age of two years due to immunodeficiency and progressive ataxia. Ocular telangiectasia were first observed at the age of three years, cutaneous telangiectasia were first documented at the age of six years and increased markedly over time even in non sun exposed areas. The boy suffered

from combined immunodeficiency with a hyper-IgM phenotype and received monthly i.v immunoglobulins, antibiotic and antifungal prophylaxis after recurrent bacterial and fungal invasive infections. He suffered from chronic EBV- and CMV-infection without obvious lymphoproliferation. Since early childhood chronic gastrointestinal disease with daily watery diarrhea and progressive liver disease with increased liver enzymes and hyperechogenic structure (ultrasound) was observed. There was severe failure to thrive despite enteral tube feeding. Furthermore, the boy suffered from joint disease mimicking juvenile polyarthritis.

Clinical findings: On admission, body weight was 16 kg, length 122 cm. Heart rate was 78 bpm, blood pressure 106/88 mmHg. ENT examination showed an anterior and posterior nasal bleeding. The intranasal mucosa showed several spots of bleeding and telangiectasia (figure 1, A-C). Hb was 8.9 g/dl (one week prior to admission Hb was 10.4 g/dl), platelet counts and plasmatic coagulation parameters (INT, PTT) were within normal ranges. Liver function tests were abnormal with increased ALT, AST and gGT. Successive ultrasound examinations showed normal liver size and increased echogenicity with no signs of primary sclerosing cholangitis or portal hypertension. Studies for lamblasis and cryptosporidiosis (stool microscopy and antigen testing) were negative.

Therapy: Prior to successful laser vaporization the boy received three erythrocyte concentrate transfusions (in total 60ml/kg bodyweight) within the first three days upon admission due to uncontrollable epistaxis. The treatment with cooling packs and derivatives of Xylometazolin as well as anterior nasal packing were ineffective. A combined anterior and nasopharyngeal tamponade was necessary to stop bleeding at first. Since any manipulation caused a new bleeding spot, bipolar coagulation could not be the treatment of choice.

Corresponding to the established treatment of M. Osler we tried a vaporization with the non-contact technique using a Nd:YAG laser (Dornier MedTech, Fibertom 5100) in general anaesthesia (figure 1, D). Starting with 5 W, continuous wave mode, the power was raised to 7 W. Before vaporization and in between the mucosa was cooled with iced cotton wool to avoid deep coagulation defects. The patient left operation theatre without any tamponade. No further epistaxis occurred within the following 15 days.

During inpatient treatment the liver function deteriorated continuously. The boy developed hepatic encephalopathy and died from liver failure of unknown origin 15 days after laser vaporisation. Autopsy and liver biopsy were denied by the parents.

In conclusion, epistaxis in AT patients may be due to endonasal telangiectasia and should prompt for early ENT examination. Treatment with laser vaporization was successful in our case.

Conflict of interest: The authors declare no conflict of interest.

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Figure 1

Endoscopic findings during surgery:

A Telangiectasia within the nasal cavity; * posterior naris

B Telangiectasia, posterior naris; * Torus tubarius

C Nasal cavity with middle turbinate (*) and medial wall of the maxillary sinus

D Aspect after vaporization of telangiactasia on the middle turbinate (*)

