



Subcapsular Hematoma of The Liver Revealing Rendu-Osler Disease: About A Pediatric Case

F Z. Khay

Pediatric Hematology and Oncology Center

M. ElBaz

Pediatric Hematology and Oncology Center

J. ElHoudzi

Pediatric Hematology and Oncology Center

ABSTRACT

RENDU-OSLER's disease is a rare hereditary disease of autosomal dominant transmission, it is a condition characterized by mucocutaneous and visceral telangiectasias; often revealed by recurrent epistaxis, hemostasis can be difficult. We report the observation of a 10-year-old girl who presented with chronic abdominal pain secondary to a subcapsular hematoma of the liver due to the rupture of an arteriovenous malformation falling within the framework of Rendu-Osler

INTRODUCTION

Rendu-Osler disease or hereditary hemorrhagic telangiectasia is a hereditary vascular dysplasia due to a mutation of one of two genes: ENG (endoglin protein) or ACVRL1 (activin receptor-like kinase 1 or ALK1 protein) identified between 1994 and 1996; responsible for vascular dysplasia in the form of telangiectasia or arteriovenous malformation on the skin, mucosa, liver, CNS [1;2]. It is characterized by hemorrhagic manifestations such as recurrent epistaxis, mucocutaneous telangiectasias and visceral arteriovenous malformations (AVMs). It is an underestimated condition in children because manifestations such as telangiectasia or epistaxis can appear late in puberty or even in adulthood. Its prevalence is estimated between 1/5000 and 1/10,000 [2].

OBSERVATION

This is a 10-year-old girl from a 2nd degree consanguineous marriage with a history of uncomplicated viral hepatitis A at the age of 3 and recurrent epistaxis. The patient who consulted for abdominal pain evolving for 1 month and not improved by symptomatic treatment; what motivated the mother to bring her daughter back to a traditional healer where an abdominal massage was done; the evolution was marked by the aggravation of the symptomatology with accentuation of the abdominal pain with on the clinical examination a tenderness of the right flank. The patient was admitted to the pediatric emergency room where an abdominal ultrasound was performed showing left liver lesions that could be of hydatid origin;

DISCUSSION

The disease described by Rendu (in Paris in 1896); by Osler (in Baltimore in 1901) and by Weber (in London in 1907) was renamed "hereditary hemorrhagic telangiectasia (HHT)" by Hanes in 1909. It is a rare but ubiquitous hereditary vascular disease with a frequency of about 1 /6000 births [3]. This pathology of vascular deregulation is expressed in the sense of hyper-angiogenesis. Cutaneomucosal telangiectasias are responsible for nasal and digestive hemorrhages and visceral vascular dilatations are responsible for arteriovenous shunts [4].

It is a genetic disease of autosomal dominant transmission, of variable expressiveness and penetrance. Genetically, there are at least two forms: HHT1 for which the anomaly is located on chromosome 9q33–34, the mutated gene being that of endoglin (ENG), and HHT2 for which the anomaly is located on chromosome 12 (12q13) the mutated gene being that of activin receptor like kinase 1 (ALK1). Endoglin and alK1 are both membrane receptors of the TGF- β (transforming growth factor β) system expressed on the surface of endothelial cells. TGF- β participates in the control of angiogenesis and vasculogenesis by intervening in the migration, proliferation and adhesion of endothelial cells and in the organization of the extracellular matrix. More recently,

Epistaxis is the most common manifestation of Rendu-Osler disease: it is found in more than 95% of patients. The epistaxis is recurrent; common and can be a source of chronic anemia [6].

The diagnosis is clinical and is based on several criteria (Curaçao criteria) [7]:

- The hereditary character
- Telangiectasias: they are one of the characteristic lesions of the disease and can be cutaneous or mucous membranes
- Epistaxis often the first manifestation of the disease
- Visceral arteriovenous malformations: hepatic; pulmonary; neurological and digestive

The clinical diagnosis is:

- Certain if at least 3 criteria are present
- Suspected or possible if 2 criteria are present
- Unlikely if only 1 criterion is present

Our patient had 2 out of 4 criteria: recurrent epistaxis and hepatic arteriovenous malformation

Patients are taken care of with their parents, in multidisciplinary consultation, in order to carry out an assessment of the lesions and the prevention of complications. Consultations in genetics, pediatrics with research of vascular murmur and measurement of pulsed oxygen saturation (SpO₂) and dermatology, supplemented by ENT nasofibroscopy, contrast echocardiography, possible chest CT scan (CT), cerebral magnetic resonance imaging (MRI) and abdominal ultrasound allow collegial discussion leading to planning adapted to each child [1].

First-line treatment of epistaxis is based on digital compression maneuvers and the use of haemostatic pads; resorting to techniques of selective embolization or vascular ligation by surgical way can be proposed in case of incoercible epistaxis. Simple advice should be given to

patients and parents; in particular the need to keep the nasal cavities in a humid environment [7].

Bevacizumab; anti-VEGF monoclonal antibody has been proposed in the treatment of Rendu-Osler disease. A few clinical cases or small series relating to the efficacy of this molecule used topically or systemically in epistaxis or Rendu-Osler anemia have thus been reported [8;9].

Most patients with hepatic arteriovenous malformations are asymptomatic and therefore do not require specific treatment. Treatment should be offered for symptomatic shunts and depends on the type of complication [10].

Radiological embolization of fistulas is not recommended because of the risk of hepatic or biliary necrosis; this is the case of our patient who remained in therapeutic abstention and the evolution was marked by the spontaneous progressive resorption of her subcapsular hematoma.

CONCLUSION

ROM is a multi-systemic vascular dysplasia, rare in children but probably underestimated due to the frequency of asymptomatic cases in childhood.

The early management of children carrying the family mutation, and proper information for the parents, aim to avoid pulmonary or cerebral complications likely to occur early. Multidisciplinary consultations allow families a complete assessment in a single place and time.

The prognosis of the disease, markedly improved by the detection measures, depends on the intensity of the epistaxis and any visceral manifestations, the morbidity of which is high; mortality is not yet measured.

The use of modulators of angiogenesis represents a promising therapeutic approach.

References

1. C. fagnou Rendu–Osler disease in children Hereditary hemorrhagic telangiectasia in children Volume 4, Issue 3, September 2021
2. Maaloul, I.; Aloulou, H.; Fourati, H.; Sfaihi, L.; Chabchoub, I.; Kamoun, T.; Mnif, Z.; Hachicha, M. (2014). Rendu-Osler-Weber disease: about a new pediatric observation. Archives of Pediatrics, 21(7), 768–771. doi: 10.1016/j.arcped.2014.04.021
3. Plauchu H, de Chadarevian JP, Bideau A, et al. Age-related clinical profile of hereditary hemorrhagic telangiectasia in an epidemiologically recruited population. Am J Med Genet 1989; 32:291–7
4. Shovlin CL. Hereditary haemorrhagic telangiectasia: pathophysiology, diagnosis and treatment. Blood Rev 2010; 24:203–19
5. V. Cottin, A.-S. Blanchet, J.-F. Cordier; Pulmonary Vascular Manifestations of Rendu-Osler Disease: Screening, Diagnosis and Treatment Pneumology Department, Reference Center for Orphan Pulmonary Diseases, Louis Pradel Hospital, Lyon I University, Rendu-Osler Disease Research Network, UMR 754, IFR 128 Lyon, France

6. Zalagh, M.; Errami, N.; Bouaity, B.; Hemmaoui, B.; Itoua, W.; Benariba, F. (2011). Severe epistaxis from Rendu-Osler-Weber disease. *Journal of Stomatology and Maxillofacial Surgery*, 112(5), 310–312.doi: 10.1016/j.stomax.2011.08.016
7. Guidelines of the French Society of Otorhinolaryngology (SFORL) (short version). Specific treatment of epistaxis in Rendu-Osler-Weber disease L.Robard, J. Michel, V. Prulière Escabasse, E. Bequignon, B. Vérillaud, O. Malard, L. Crampette, The SFORL Work-Group, O. Malard, L. Crampette, M. Achache, MY Alaoui Lamrani , L. Ardillon, E. Babin, C. Bal Dit Sollier, E. Bequignon, M. Borsik, L. Castillo, A. Coste, C. Debry, P. Dessi, L. Drouet, X. Dufour, S. Dupuis-Girod, F. Faure, P. Gallet, R. Guldman, E. Houdart, R. Jankowski, F. Jegoux, S. Leble, J. Michel, G. Mortuaire, E. Mouchon, C. Page, V. Pruliere Escabasse, L. Robard, A. Roux, JP Saint Maurice, G. Sarlon, V. Strunski, V. Trevillot, B. Verillaud, P. Vironneau
8. gaillard s, dupuis-girod s, Boutitie f, et al. tranexamic acid for epistaxis in hereditary hemorrhagic telangiectasia patients: a european cross-over controlled trial in a rare disease. *J thromb haemost* 2014; 12:1494-502.
9. chen st, karnezis t, davidson tm. Safety of intranasal Bevacizumab (avastin) treatment in patients with hereditary hemorrhagic telangiectasia-associated epistaxis. *laryngoscope* 2011; 121:644-6.
10. Liver damage in Rendu-Osler disease: about a case and review of the literature Hanen Loukil,Mouna Snoussi,Hela Fourati,Faten Frikha,Raida Ben Salah,Moez Jallouli,Sameh Marzouk,Zeineb Mnif, AndZouhir Bahloul