Published online May 2, 2011
PEDIATRICS (doi:10.1542/peds.2010-2366)

CASE REPORTS

Symptomatic Liver Involvement in Neonatal Hereditary Hemorrhagic Telangiectasia

Suhail Al-Saleh, MBBS, MSc, FRCPC[a,b,c,d], Philip R. John, MBChB, DCH, FRCR, FRCPC[a,e], Michelle Letarte, PhD[a,f], Marie E. Faughnan, MD, MSc[a,g,h], Jaques Belik, MD, FRCPC[c,i], Felix Ratjen, MD, PhD, FRCPC[a,b,c]

[a]University of Toronto, Toronto, Ontario, Canada;
[b]Division of Respiratory Medicine, Department of Pediatrics,
[c]Physiology and Experimental Medicine, Research Institute,
[e]Department of Diagnostic Imaging,
[f]Molecular Structure and Function, and
[i]Division of Neonatology, Hospital for Sick Children, Toronto, Ontario, Canada;
[d]King Saud bin Abdulaziz University for Health Sciences, Riyadh, Saudi Arabia; and
[g]Department of Medicine and
[h]Li Ka Shing Knowledge Institute, St Michael's Hospital, Toronto, Ontario, Canada

High-flow hepatic vascular anomalies with arteriovenous shunting commonly manifest during the neonatal period with signs and symptoms of congestive heart failure, but to our knowledge, they have never been described in patients with hereditary hemorrhagic telangiectasia (HHT). We report here our experience with 3 patients with hepatic arteriovenous malformations (AVMs) who presented with symptoms of high-output congestive heart failure during the neonatal period and were subsequently diagnosed with HHT. Imaging showed large hypervascular lesions and multiple hepatic arteriovenous shunts that differentiated these lesions from liver hemangiomas. Transcatheter embolization was performed in all cases. One infant died of sepsis shortly after embolization; follow-up at the age of 2.5 years of the surviving infants revealed involution of the vascular lesions and no evidence of symptom recurrence. We conclude that severe symptoms related to hepatic AVMs in HHT can occur in the neonatal period and that HHT should therefore be included in the differential diagnosis of symptomatic neonatal hepatic vascular malformations. Imaging plays a key role in differentiating hepatic AVMs from hemangiomas, because the latter require additional pharmacologic treatments. Early transcatheter embolization seems to be effective, but long-term outcomes still need to be assessed.

Key Words: hereditary hemorrhagic telangiectasia • HHT • genetics • Rendu-Osler-Weber • arteriovenous malformations • visceral • liver • children

Abbreviations: HHT = hereditary hemorrhagic telangiectasia • CHF = congestive heart failure • AVM = arteriovenous malformation • *ALK1* = activin receptor-like kinase 1

Accepted Jan 26, 2011.