Vascular Anomalies:
Diagnosis and Management

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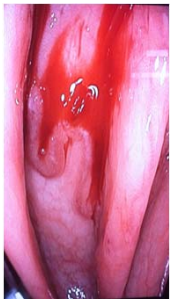
Boston Children's Hospital

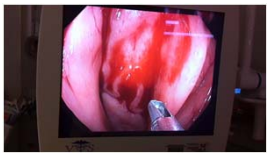
HARVARD MEDICAL SCHOOL

Disclosure

In the past 12 months, I have no relevant financial relationships with the manufacturers of any commercial products or providers of commercial services discussed in this CME activity.

What is this lesion?





Learning Objectives

- Define "vascular anomalies"
- Learn approach to classification
- Review diagnosis and management of specific lesions
  - Alimentary tract (exclude hepatic lesions)

Definition of vascular anomalies

- Morphologically and biologically diverse group of abnormalities of vascular channels
- Often congenital but grow or change with time
- May involve single or a combination of vessel types: capillary, venous, arterial, and lymphatic
- May represent isolated lesions or part of multiorgan system condition

Classification

- Primary classification
  - Tumors
  - Malformations
- Based on biological behavior, clinical behavior, and physical endothelial chararacteristics
- Predicts likely response to antiproliferative drugs vs surgical or other ablative therapy

* Mulliken JB and Glowacki J. Plast Reconstr Surg 1982;69:412

Classification for Vascular Anomalies

International Society for the Study of Vascular Anomalies (ISSVA)

(Approved at the 20th ISSVA Workshop, Melbourne, April 2014)

Vascular anomalies				
Vascular tumors	**Vascular malformations**			
	Simple	**Combined** °	**of major named vessels**	**associated with other anomalies**
Benign Infantile hemangioma Congenital hemangioma Locally aggressive or borderline Kaposiform hemangioendothelioma (KHE) Malignant angiosarcoma	Capillary malformations port wine stain Lymphatic malformations Venous malformations Blue rubber bleb nevus Arteriovenous malformations* Arteriovenous fistula*	CVM, CLM LVM, CLVM CAVM* CLAVM* others	e.g. aorta	e.g. Klippel-Trenaunay Syndrome (KTS) CLVM with limb overgrowth

° defined as two or more vascular malformations found in one lesion

• high-flow lesions

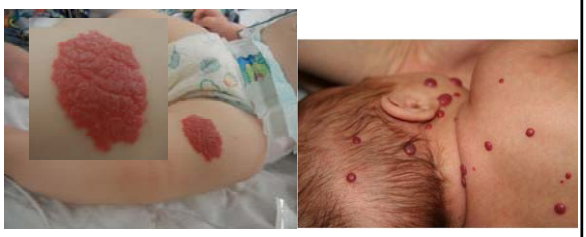
Clinical GI Presentations

- Gastrointestinal bleeding
  - Overt
  - Occult with chronic anemia
- Abdominal pain, intestinal obstruction
- Intestinal malabsorption, PLE, diarrhea, hypoalbuminemia, ascites

Hemangioma

- Most common tumor of infancy and childhood
- Incidence (neonates) 1-2.6%
  - <1 kg premi 23%
  - <1.5 kg 15%
- Gender = 3:1 F:M
- Distribution
  - Solitary 80%
  - Multifocal 20%
    - Often with intracranial or visceral involvement
- Cutaneous locations
  - Head and neck 60%
  - Trunk 25%
  - Extremity 15%
- Extra-cutaneous locations
  - Likely if > 5 cutaneous lesions
  - Any, but most common are liver, lung, brain, intestine
- Three growth phases
  - Proliferative neonate
  - Involution (early) 1- 5 y
  - Involution (late) > 5 y

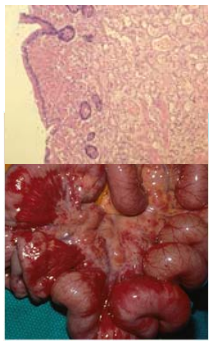
Hemangioma



Solitary Multifocal

Intestinal Hemangioma

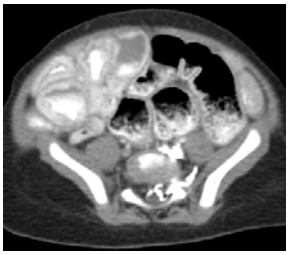
- 16 children in BCH VAC database 1998-2012
- 14F: 2M
- 14 (88%) presented with GI bleeding in first 4 mos of life
- 2 bowel perforations



Soukoulis et al (manuscript submitted)

Intestinal Hemangioma

- Diagnosis (n)
  - Laparotomy 8
  - CT/MRI/US 5
  - Angiogram 1
  - Colonoscopy 1
  - WCE 1
  - EGD/colon (-) 8/9
- Involved site
  - SB 15
  - Colon 1
  - Cutaneous 9 (56%)



CT with enhancing bowel wall thickening

Intestinal Hemangioma

- 7/16 with congenital abnormalities
  - Pierre Robin sequence
  - Short bowel and malrot
  - VSD and pulm HTN
  - Tetralogy of Fallot
  - Cleft lip/palate, micrognathia
  - Tailgut cyst
  - PHACES association (n=1)
    - Posterior fossa tumor
    - Hemangioma of face
    - Arterial abnormality
    - Cardiac anomaly
    - Eye abnormality
    - Sternal defects

Regional facial hemangioma

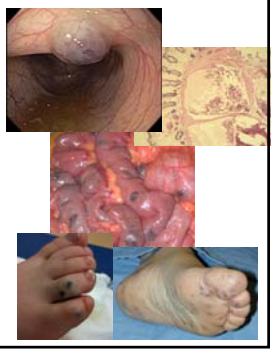
Intestinal Hemangioma: Treatment

- Therapeutic options
  - Propranolol (2 mg/kg/day)
    - Should be first choice
  - Corticosteroids (2-4 mg/kg/day)
    - Well established efficacy
  - Vincristine (low dose)
  - Combination drug therapy
  - Bowel resection (last resort)
- Duration of therapy
  - Usually 6-9 months, but sometimes longer
  - Major endpoint is cessation of bleeding

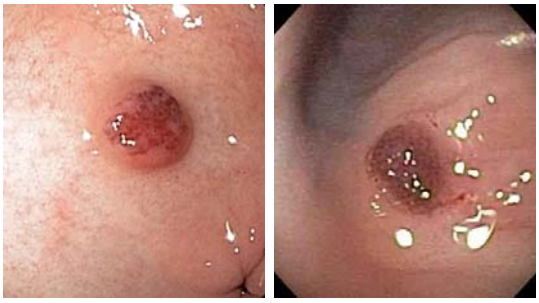
Izadpanah A et al Plast Reconstr Surg 2013;131:601-13

Blue Rubber Bleb Nevus Syndrome (BRBNS)

- Multifocal polypoid VM
  - "cavernous hemangioma"
- Incidence unknown
- Somatic mutation in TIE2 gene, chromosome 9p21.2
- Predominant skin and intestine involvement
- Presents with GI bleeding in early childhood
- Diagnosis by endoscopy



Beware of endoscopic mimicry!



BRBNS

- Soft tissue lesions detected by labeled RBC scan
- Intussusception of small bowel may be asymptomatic or present with intermittent pain or obstruction

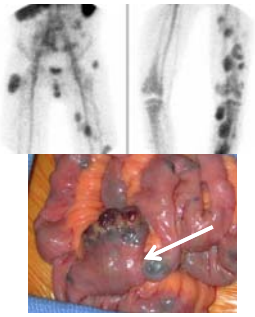
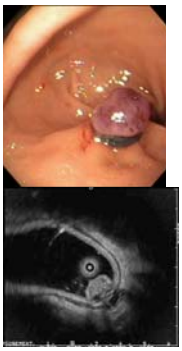


Photo courtesy of Steven Fishman, MD

BRBNS: Treatment

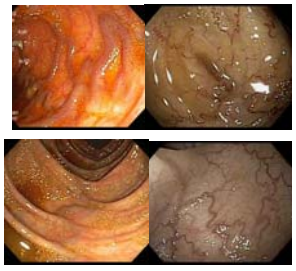
- Surgical excision is preferred definitive therapy
  - May be supplemented by endoscopic ligation/polypectomy
- Limited role for antiproliferative drugs
  - Delay surgical intervention in young children
  - Rescue therapy for poor surgical candidates
  - Sirolimus is current drug of choice



Fishman SJ et al. Ann Surg 2005;241:523-8
Yuksekkaya H, et al. Pediatrics 2012;129:e1-e5

Reticular Venous Malformation

- Superficial VM with a tortuous variceal or net-like pattern
- Often involves entire length of colon and distal ileum
- May include other SB
- Incidence is unknown
- Usually sporadic, rarely hereditary
  - autosomal dominant familial colonic varices
  - Turner's syndrome



Familial varices Turner's syndrome

Reticular VM: Treatment

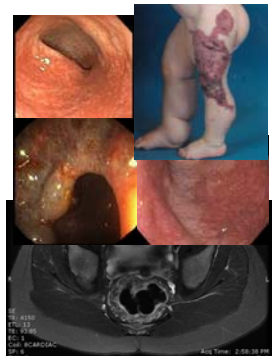
- Reduce severity of bleeding episodes with antibrinolytic or clot stabilizing drugs, eg. tranexamic acid (Lysteda) or aminocaproic acid (Amicar)
- Endoscopic sclerotherapy or thermal coagulation/ablation, eg. argon plasma coagulation
- Colectomy or segmental bowel resection

APC ablation

- Start VID 003 at -08:17

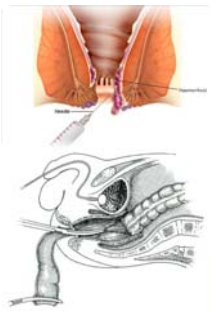
Segmental Colonic VM

- Endoscopic appearance
  - Diffuse erythema, flat or slightly raised texture, absent branching vessel pattern
- CT or MRI shows wall thickening due to transmural infiltration
- Frequent rectosigmoid distribution
- Isolated lesion, or with Klippel- Trénaunay Syndrome



Segmental Rectosigmoid VM: Therapy

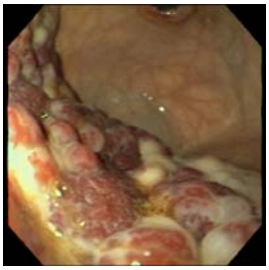
- Transanal sclerotherapy
- Partial colectomy with pull-through anastomosis



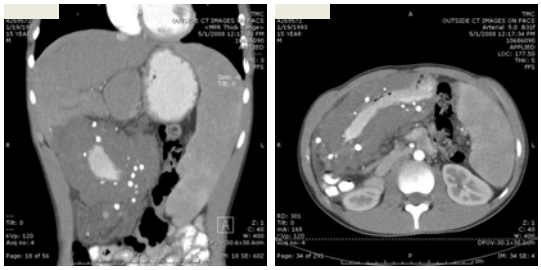
Fishman SJ et al. J Pediatr Surg 2000;35:982-4
Spitz L, Coran AG, eds. Operative Pediatric Surgery. 7th ed. Boca Raton, Fl: CRC Press; 2013

Segmental Lobular VM

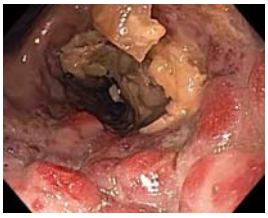
- Raised lobular surface
- Significant mass effect
- Focal
- Transmural
- Sporadic
- Treated by surgical resection



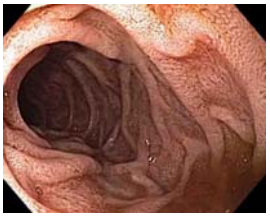
What anatomy is involved?



Valuable endoscopic Mapping!



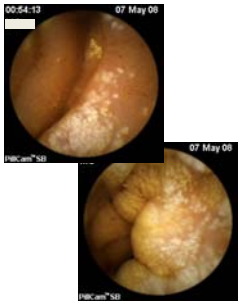
Distal stomach



Duodenum

Intestinal Lymphangiectasia

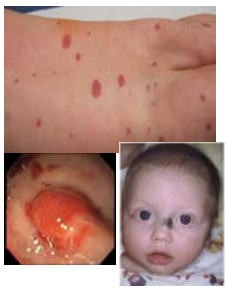
- Predominantly small bowel involvement
- Essential role of wireless capsule endoscopy
- White-tipped thick villi
- Patchy multifocal or continuous
- Malabsorption
  - Protein-losing enteropathy
  - Fat malabsorption
- Treated by reduced long chain fats, MCT oil, albumin infusions, octreotide, bowel resection
- Potential role for lymphangiography to look for disrupted thoracic duct
  - Microsurgical reconstruction?



Provisionally Unclassified Vascular Anomalies

Cutaneovisceral Angiomatosis with Thrombocytopenia (CAT)

- Also called "multifocal lymphangioendotheliomatosis with thrombocytopenia (MLT)"*
- Multiple red, purple, or brown cutaneous lesions
- Hemangioma-like gastrointestinal and pulmonary lesions
  - Also liver, spleen, muscle, bone, brain, retina
- Treat with corticosteroids, vincristine, thalidomide to control bleeding
- Partial regression over months to years but long-term outcome unknown



*North et al, Arch Dermatol 2004;140:599
Prasad et al, Pediatr Dev Pathol 2005;8:407-19

Summary

- Careful gross anatomic, endoscopic, and microscopic description of GI vascular anomalies is essential for correct classification
- Classification and distribution of lesion(s) determines most effective treatment
- A coordinated multidisciplinary approach facilitates optimal management and advances scientific understanding